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(54) Title: NUCLEIC ACIDS CONTAINING SINGLE NUCLEOTIDE POLYMORPHISMS AND METHODS OF USE THEREOF

(57) Abstract: The invention provides nucleic acids containing single-nucleotide polymorphisms identified for transcribed human sequences, as well as methods of using the nucleic acids.

# NUCLEIC ACIDS CONTAINING SINGLE NUCLEOTIDE POLYMORPHISMS AND METHODS OF USE THEREOF

## FIELD OF THE INVENTION

The invention relates generally to nucleic acids and polypeptides and in particular to  
5 the identification of human single nucleotide polymorphisms based on at least one gene product that was not previously described.

## BACKGROUND OF THE INVENTION

Sequence polymorphism-based analysis of nucleic acid is generally based on alterations in nucleic acid sequences between related individuals. This analysis has been  
10 widely used in a variety of genetic, diagnostic, and forensic applications. For example, polymorphism analyses are used in identity and paternity analysis, and in genetic mapping studies.

Several different types of polymorphisms in nucleic acid have been described. One such type of variation is a restriction fragment length polymorphism (RFLP). RFLPs can  
15 create or delete a recognition sequence for a restriction endonuclease in one nucleic acid relative to a second nucleic acid. The result of the variation is in an alteration the relative length of restriction enzyme generated DNA fragments in the two nucleic acids.

Other polymorphisms take the form of short tandem repeats (STR) sequences, which are also referred to as variable numbers of tandem repeat (VNTR) sequences. STR sequences  
20 typically include tandem repeats of 2, 3, or 4 nucleotide sequences that are present in a nucleic acid from one individual but absent from a second, related individual at the corresponding genomic location.

Other polymorphisms take the form of single nucleotide variations, termed single nucleotide polymorphisms (SNPs), between individuals. A SNP can, in some instances, be  
25 referred to as a "cSNP" to denote that the nucleotide sequence containing the SNP originates as a cDNA.

SNPs can arise in several ways. A single nucleotide polymorphism may arise due to a substitution of one nucleotide for another at the polymorphic site. Substitutions can be transitions or transversions. A transition is the replacement of one purine nucleotide by

another purine nucleotide, or one pyrimidine by another pyrimidine. A transversion is the replacement of a purine by a pyrimidine, or the converse.

Single nucleotide polymorphisms can also arise from a deletion of a nucleotide or an insertion of a nucleotide relative to a reference allele. Thus, the polymorphic site is a site at 5 which one allele bears a gap with respect to a single nucleotide in another allele. Some SNPs occur within, or near genes. One such class includes SNPs falling within regions of genes encoding for a polypeptide product. These SNPs may result in an alteration of the amino acid sequence of the polypeptide product and give rise to the expression of a defective or other variant protein. Such variant products can, in some cases result in a pathological condition, 10 e.g., genetic disease. Examples of genes in which a polymorphism within a coding sequence gives rise to genetic disease include sickle cell anemia and cystic fibrosis. Other SNPs do not result in alteration of the polypeptide product. Of course, SNPs can also occur in noncoding regions of genes.

SNPs tend to occur with great frequency and are spaced uniformly throughout the 15 genome. The frequency and uniformity of SNPs means that there is a greater probability that such a polymorphism will be found in close proximity to a genetic locus of interest.

## SUMMARY OF THE INVENTION

The invention is based in part on the discovery of single nucleotide polymorphisms (SNPs) in regions of human DNA.

20 Accordingly, in one aspect, the invention provides nucleic acid sequences comprising nucleic acid segments of both publicly known and novel genes, including the polymorphic site. The segments can be DNA or RNA, and can be single- or double-stranded. Preferred segments include a biallelic polymorphic site.

25 The invention further provides allele-specific oligonucleotides that hybridize to a segment of a fragment shown in Table 1, column 4, or its complement. These oligonucleotides can be probes or primers. Also provided are isolated nucleic acids comprising a sequence shown in Table 1, column 4, in which the polymorphic site within the sequence is occupied by a base other than the reference bases shown in Table 1, columns 5 and 6.

The invention further provides a method of analyzing a nucleic acid from an individual. The method determines which base is present at any one of the polymorphic sites shown in Table 1. Optionally, a set of bases occupying a set of polymorphic sites shown in Table 1 is determined. This type of analysis can be performed on a number of individuals, 5 who are tested for the presence of a disease phenotype.

In another aspect, the invention provides an isolated polynucleotide which includes one or more of the SNPs described herein. The polynucleotide can be, *e.g.*, a nucleotide sequence which includes one or more of the polymorphic sequences shown in Table 1 and which includes a polymorphic sequence, or a fragment of the polymorphic sequence, as long 10 as it includes the polymorphic site. The polynucleotide may alternatively contain a nucleotide sequence which includes a sequence complementary to one or more of these sequences, or a fragment of the complementary nucleotide sequence, provided that the fragment includes a polymorphic site in the polymorphic sequence.

The polynucleotide can be, *e.g.*, DNA or RNA, and can be between about 10 and 15 about 100 nucleotides, *e.g.* 10-90, 10-75, 10-51, 10-40, or 10-30, nucleotides in length.

In preferred embodiments, the polymorphic site in the polymorphic sequence includes a nucleotide other than the nucleotide listed in Table 1, column 5 for the polymorphic sequence, *e.g.*, the polymorphic site includes the nucleotide listed in Table 1, column 6 for the polymorphic sequence.

20 In other embodiments, the complement of the polymorphic site includes a nucleotide other than the complement of the nucleotide listed in Table 1, column 5 for the complement of the polymorphic sequence, *e.g.*, the complement of the nucleotide listed in Table 1, column 6 for the polymorphic sequence.

25 In some embodiments, the polymorphic sequence is associated with a polypeptide related to one of the protein families disclosed herein. For example, the nucleic acid may be associated with a polypeptide related to angiopoietin, 4-hydroxybutyrate dehydrogenase, or any of the other proteins identified in Table 1, column 10.

30 In another aspect, the invention provides an isolated allele-specific oligonucleotide that hybridizes to a first polynucleotide containing a polymorphic site. The first polynucleotide can be, *e.g.*, a nucleotide sequence comprising one or more polymorphic

sequences recited in Table 1, provided that the polymorphic sequence includes a nucleotide other than the nucleotide recited in Table 1, column 5 for the polymorphic sequence.

Alternatively, the first polynucleotide can be a nucleotide sequence that is a fragment of the polymorphic sequence, provided that the fragment includes a polymorphic site in the

5 polymorphic sequence, or a complementary nucleotide sequence which includes a sequence complementary to one or more polymorphic sequences in Table 1, provided that the complementary nucleotide sequence includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5. The first polynucleotide may in addition include a nucleotide sequence that is a fragment of the complementary sequence, provided that the

10 fragment includes a polymorphic site in the polymorphic sequence.

In some embodiments, the oligonucleotide does not hybridize under stringent conditions to a second polynucleotide. The second polynucleotide can be, e.g., (a) a nucleotide sequence comprising one or more polymorphic sequences in Table 1, wherein the polymorphic sequence includes the nucleotide listed in Table 1, column 5 for the

15 polymorphic sequence; (b) a nucleotide sequence that is a fragment of any of the polymorphic sequences; (c) a complementary nucleotide sequence including a sequence complementary to one or more polymorphic sequences disclosed herein in Table 1; and (d) a nucleotide sequence that is a fragment of the complementary sequence, provided that the fragment includes a polymorphic site in the polymorphic sequence.

20 The oligonucleotide can be, e.g., between about 10 and about 100 bases in length. In some embodiments, the oligonucleotide is between about 10 and 75 bases, 10 and 51 bases, 10 and about 40 bases, or about 15 and 30 bases in length.

The invention also provides a method of detecting a polymorphic site in a nucleic acid. The method includes contacting the nucleic acid with an oligonucleotide that hybridizes  
25 to a polymorphic sequence selected shown in Table 1, or its complement, provided that the polymorphic sequence includes a nucleotide other than the nucleotide recited in Table 1, column 5 for the polymorphic sequence, or the complement includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5. The method also includes determining whether the nucleic acid and the oligonucleotide hybridize. Hybridization of the  
30 oligonucleotide to the nucleic acid sequence indicates the presence of the polymorphic site in the nucleic acid.

In preferred embodiments, the oligonucleotide does not hybridize to the polymorphic sequence when the polymorphic sequence includes the nucleotide recited in Table 1, column 5 for the polymorphic sequence, or when the complement of the polymorphic sequence includes the complement of the nucleotide recited in Table 1, column 5 for the polymorphic sequence.

The oligonucleotide can be, *e.g.*, between about 10 and about 100 bases in length. In some embodiments, the oligonucleotide is between about 10 and 75 bases, 10 and 51 bases, 10 and about 40 bases, or about 15 and 30 bases in length.

In some embodiments, the polymorphic sequence identified by the oligonucleotide is associated with a nucleic acid encoding polypeptide related to one of the protein families disclosed herein. the polymorphic sequence is associated with a polypeptide related to one of the protein families disclosed herein. For example, the nucleic acid may be associated with a polypeptide related to angiopoietin, 4-hydroxybutyrate dehydrogenase, or any of the other proteins identified in Table 1, column 10.

In a further aspect, the invention provides a method of determining the relatedness of a first and second nucleic acid. The method includes providing a first nucleic acid and a second nucleic acid and contacting the first nucleic acid and the second nucleic acid with an oligonucleotide that hybridizes to a polymorphic sequence selected disclosed in Table 1, or its complement, provided that the polymorphic sequence includes a nucleotide other than the nucleotide recited in Table 1, column 5 for the polymorphic sequence, or the complement includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5. The method also includes determining whether the first nucleic acid and the second nucleic acid hybridize to the oligonucleotide, and comparing hybridization of the first and second nucleic acids to the oligonucleotide. Hybridization of first and second nucleic acids to the nucleic acid indicates the first and second subjects are related.

In preferred embodiments, the oligonucleotide does not hybridize to the polymorphic sequence when the polymorphic sequence includes the nucleotide recited in Table 1, column 5 for the polymorphic sequence, or when the complement of the polymorphic sequence includes the complement of the nucleotide recited in Table 1, column 5 column for the polymorphic sequence.

The oligonucleotide can be, *e.g.*, between about 10 and about 100 bases in length. In some embodiments, the oligonucleotide is between about 10 and 75 bases, 10 and 51 bases, 10 and about 40 bases, or about 15 and 30 bases in length.

The method can be used in a variety of applications. For example, the first nucleic acid may be isolated from physical evidence gathered at a crime scene, and the second nucleic acid may be obtained is a person suspected of having committed the crime. Matching the two nucleic acids using the method can establishing whether the physical evidence originated from the person.

In another example, the first sample may be from a human male suspected of being the father of a child and the second sample may be from a child. Establishing a match using the described method can establishing whether the male is the father of the child.

In another aspect, the method includes determining if a sequence polymorphism is the present in a subject, such as a human. The method includes providing a nucleic acid from the subject and contacting the nucleic acid with an oligonucleotide that hybridizes to a polymorphic sequence disclosed in Table 1, or its complement, provided that the polymorphic sequence includes a nucleotide other than the nucleotide recited in Table 1, column 5 for the polymorphic sequence, or the complement includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5. Hybridization between the nucleic acid and the oligonucleotide is then determined. Hybridization of the oligonucleotide to the nucleic acid sequence indicates the presence of the polymorphism in said subject.

In another aspect, the invention provides an isolated polypeptide comprising a polymorphic site at one or more amino acid residues, and wherein the protein is encoded by a polynucleotide including one of the polymorphic sequences in Table 1, or their complement, provided that the polymorphic sequence includes a nucleotide other than the nucleotide recited in Table 1, column 5 for the polymorphic sequence, or the complement includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5.

The polypeptide can be, *e.g.*, related to one of the protein families disclosed herein. For example, polypeptide can be related to angiopoietin, 4-hydroxybutyrate dehydrogenase, ATP-dependent RNA helicase, MHC Class I histocompatibility antigen, or phosphoglycerate kinase.

In some embodiments, the polypeptide is translated in the same open reading frame as is a wild type protein whose amino acid sequence is identical to the amino acid sequence of the polymorphic protein except at the site of the polymorphism.

In some embodiments, the polypeptide encoded by the polymorphic sequence, or its 5 complement, includes the nucleotide listed in Table 1, column 6 for the polymorphic sequence, or the complement includes the complement of the nucleotide listed in Table 1, column 6.

The invention also provides an antibody that binds specifically to a polypeptide encoded by a polynucleotide comprising a nucleotide sequence encoded by a polynucleotide 10 including one or more of the polymorphic sequences in Table 1, or its complement. The polymorphic sequence includes a nucleotide other than the nucleotide recited in Table 1, column 5 for the polymorphic sequence, or the complement includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5.

In some embodiments, the antibody binds specifically to a polypeptide encoded by a 15 polymorphic sequence which includes the nucleotide listed in Table 1, column 6 for the polymorphic sequence.

Preferably, the antibody does not bind specifically to a polypeptide encoded by a polymorphic sequence which includes the nucleotide listed in Table 1, column 5 for the polymorphic sequence.

20 The invention further provides a method of detecting the presence of a polypeptide having one or more amino acid residue polymorphisms in a subject. The method includes providing a protein sample from the subject and contacting the sample with the above-described antibody under conditions that allow for the formation of antibody-antigen complexes. The antibody-antigen complexes are then detected. The presence of the 25 complexes indicates the presence of the polypeptide.

The invention also provides a method of treating a subject suffering from, at risk for, or suspected of, suffering from a pathology ascribed to the presence of a sequence polymorphism in a subject, *e.g.*, a human, non-human primate, cat, dog, rat, mouse, cow, pig, goat, or rabbit. The method includes providing a subject suffering from a pathology 30 associated with aberrant expression of a first nucleic acid comprising a polymorphic sequence

shown in Table 1, or its complement, and treating the subject by administering to the subject an effective dose of a therapeutic agent. Aberrant expression can include qualitative alterations in expression of a gene, *e.g.*, expression of a gene encoding a polypeptide having an altered amino acid sequence with respect to its wild-type counterpart. Qualitatively different polypeptides can include, shorter, longer, or altered polypeptides relative to the amino acid sequence of the wild-type polypeptide. Aberrant expression can also include quantitative alterations in expression of a gene. Examples of quantitative alterations in gene expression include lower or higher levels of expression of the gene relative to its wild-type counterpart, or alterations in the temporal or tissue-specific expression pattern of a gene.

Finally, aberrant expression may also include a combination of qualitative and quantitative alterations in gene expression.

The therapeutic agent can include, *e.g.*, second nucleic acid comprising the polymorphic sequence, provided that the second nucleic acid comprises the nucleotide present in the wild type allele. In some embodiments, the second nucleic acid sequence comprises a polymorphic sequence which includes nucleotide listed in Table 1, column 5 for the polymorphic sequence.

Alternatively, the therapeutic agent can be a polypeptide encoded by a polynucleotide comprising polymorphic sequence shown in Table 1, or by a polynucleotide comprising a nucleotide sequence that is complementary to any one of the polymorphic sequences, provided that the polymorphic sequence includes the nucleotide listed in Table 1, column 6 for the polymorphic sequence.

The therapeutic agent may further include an antibody as herein described, or an oligonucleotide comprising a polymorphic sequence shown in Table 1, or by a polynucleotide comprising a nucleotide sequence that is complementary to any one the polymorphic sequences, provided that the polymorphic sequence includes the nucleotide listed in Table 1, column 6 for the polymorphic sequence,

In another aspect, the invention provides an oligonucleotide array comprising one or more oligonucleotides hybridizing to a first polynucleotide at a polymorphic site encompassed therein. The first polynucleotide can be, *e.g.*, a nucleotide sequence comprising one or more polymorphic sequences shown in Table 1; a nucleotide sequence that is a fragment of any of the nucleotide sequence, provided that the fragment includes a

polymorphic site in the polymorphic sequence; a complementary nucleotide sequence comprising a sequence complementary to one or more of the polymorphic sequences; or a nucleotide sequence that is a fragment of the complementary sequence, provided that the fragment includes a polymorphic site in the polymorphic sequence.

5 In preferred embodiments, the array comprises 10; 100; 1,000; 10,000; 100,000 or more oligonucleotides.

The invention also provides a kit comprising one or more of the herein-described nucleic acids. The kit can include, *e.g.*, polynucleotide which includes one or more of the SNPs described herein. The polynucleotide can be, *e.g.*, a nucleotide sequence which 10 includes one or more of the polymorphic sequences shown in Table 1, and which includes a polymorphic sequence, or a fragment of the polymorphic sequence, as long as it includes the polymorphic site. The polynucleotide may alternatively contain a nucleotide sequence which includes a sequence complementary to one or more of the sequences, or a fragment of the complementary nucleotide sequence, provided that the fragment includes a polymorphic 15 site in the polymorphic sequence.

Alternatively, or in addition, the kit can include the invention provides an isolated allele-specific oligonucleotide that hybridizes to a first polynucleotide containing a polymorphic site. The first polynucleotide can be, *e.g.*, a nucleotide sequence comprising one or more polymorphic sequences shown in Table 1, provided that the polymorphic sequence 20 includes a nucleotide other than the nucleotide recited in Table 1, column 5 for the polymorphic sequence. Alternatively, the first polynucleotide can be a nucleotide sequence that is a fragment of the polymorphic sequence, provided that the fragment includes a polymorphic site in the polymorphic sequence, or a complementary nucleotide sequence which includes a sequence complementary to one or more polymorphic sequences shown in 25 Table 1, provided that the complementary nucleotide sequence includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 6. The first polynucleotide may in addition include a nucleotide sequence that is a fragment of the complementary sequence, provided that the fragment includes a polymorphic site in the polymorphic sequence.

**BRIEF DESCRIPTION OF THE DRAWING**

FIG. 1 illustrates an example of the way in which SNP sites were identified in the present invention.

Unless otherwise defined, all technical and scientific terms used herein have the same meaning as commonly understood by one of ordinary skill in the art to which this invention belongs. Although methods and materials similar or equivalent to those described herein can be used in the practice or testing of the present invention, suitable methods and materials are described below. All publications, patent applications, patents, and other references mentioned herein are incorporated by reference in their entirety. In the case of conflict, the present specification, including definitions, will control. In addition, the materials, methods, and examples are illustrative only and not intended to be limiting.

Other features and advantages of the invention will be apparent from the following detailed description and claims.

**DETAILED DESCRIPTION OF THE INVENTION**

The invention provides human SNPs in sequences which are transcribed, *i.e.*, are cSNPs. Many SNPs have been identified in genes related to polypeptides of known function. If desired, SNPs associated with various polypeptides can be used together. For example, SNPs can be grouped according to whether they are derived from a nucleic acid encoding a polypeptide related to particular protein family or involved in a particular function.

Similarly, SNPs can be grouped according to the functions played by their gene products. Such functions include, structural proteins, proteins from which associated with metabolic pathways fatty acid metabolism, glycolysis, intermediary metabolism, calcium metabolism, proteases, and amino acid metabolism, etc. Specifically, the present invention provides a large number of human cSNP's based on at least one gene product that has not been previously identified. In contrast, and as defined specifically in the following paragraph, the cSNP's involve nucleic acid sequences that are assembled from at least one known sequence.

The present invention describes 651 distinct polymorphic sites, which are summarized in Table 1. Raw traces underlying sequence data were drawn from public databases and from the proprietary database of the Assignee of the present invention. The sequences were obtained by calling the bases from these traces, and included assigning "Phred" quality scores

for each called base. For each allelic set, at the polynucleotide level, four or more nucleotide sequences were identified having at least partial overlap with one another.

As illustrated in FIG. 1, these four or more sequences could be clustered and assembled to make a consensus contig that included an ORF. In this way, the inventors  
5 found that the assembled contigs defined associated sets of two, or possibly more than two, alleles defined by a SNP at a particular polymorphic site. In order to be confirmed as a SNP site, the nucleotide change from the consensus sequence had to occur in at least two individual sequences, and had to have a "Phred" score of 23 or higher at the site of the presumed SNP. Furthermore, in a window of 5 bases on either side of the SNP, no more than  
10 50% mismatching with the consensus sequence was allowed. In the assembly leading to each of the contigs defining the allelic set, the SNP alleles occur in polynucleotides found in public databases.

It was found that the assembled contigs defined associated sets of two, or possibly more than two, alleles defined by an SNP at a particular polymorphic site. These associations  
15 were not previously known.

At the level of translation of an ORF contained in the contigs, allelic sets were identified in which one allele defines a known polypeptide sequence that includes the polymorphic site and another polypeptide allele is not previously known. Then, various associations of alleles are possible. For example, it is possible that an allelic pair is defined  
20 in a noncoding region of the contig containing an ORF. In such cases the inventors believe that the invention resides in the recognition of the allelic pair; this association has not heretofore been made.

Alternatively, sets of allelic contigs may exist in which the polymorphic site is within an ORF, but does not result in an amino acid change among the allelic polypeptides. Thus, in  
25 another embodiment, the polymorphic site resides within an ORF and results in an amino acid change, or a frameshift, among the alleles of the allelic set. In the sets of gene products that fall within this group, at least one of the alleles at the polypeptide level is a known protein. At least one of the remaining allele or alleles in the set, carrying a variant amino acid at the polymorphic site, is a novel polypeptide not heretofore known. The invention resides  
30 at least in the recognition of the polymorphic allele as being a variant of the known reference polypeptide.

Table 1 provides information concerning the allelic sequences. One of the sequences may be termed a reference polymorphic sequence, and the corresponding second sequence includes the variant SNP at the polymorphic site. Since the reference polypeptide sequence is already known, the Sequence Listing accompanying this application provides only the sequence of the polymorphic allele, while its SEQ ID NO is provided in the Table. A reference to the SEQ ID NO that corresponds to the translated amino acid sequence is also given. The Table includes thirteen columns that provide descriptive information for each cSNP, each of which occupies one row in the Table. The column headings, and a description of each, are given below.

SNPs disclosed in Table 1 were detected by aligning large numbers of sequences from genetically diverse sources of publicly available mRNA libraries (Clontech). Software designed specifically to look for multiple examples of variant bases differing from a consensus sequence was created and deployed. A criteria of a minimum of 2 occurrences of a sequence differing from the consensus in high quality sequence reads was used to identify an SNP.

The SNPs described herein may be useful in diagnostic kits, for DNA arrays on chips and for other uses that involve hybridization of the SNP.

Specific SNPs may have utility where a disease has already been associated with that gene. Examples of possible disease correlations between the claimed SNPs with members of the genes of each classification are listed below:

### **Amylases**

Amylase is responsible for endohydrolysis of 1,4-alpha-glucosidic linkages in oligosaccharides and polysaccharides. Variations in amylase gene may be indicative of delayed maturation and of various amylase producing neoplasms and carcinomas.

### **25 Amyloid**

The serum amyloid A (SAA) proteins comprise a family of vertebrate proteins that associate predominantly with high density lipoproteins (HDL). The synthesis of certain members of the family is greatly increased in inflammation. Prolonged elevation of plasma SAA levels, as in chronic inflammation, results in a pathological condition, called amyloidosis, which affects the liver, kidney and spleen and which is characterized by the

highly insoluble accumulation of SAA in these tissues. Amyloid selectively inhibits insulin-stimulated glucose utilization and glycogen deposition in muscle, while not affecting adipocyte glucose metabolism. Deposition of fibrillar amyloid proteins intraneuronally, as neurofibrillary tangles, extracellularly, as plaques and in blood vessels, is characteristic of both Alzheimer's disease and aged Down's syndrome. Amyloid deposition is also associated with type II diabetes mellitus.

### **Angiopoietin**

Members of the angiopoietin/fibrinogen family have been shown to stimulate the generation of new blood vessels, inhibit the generation of new blood vessels, and perform several roles in blood clotting. This generation of new blood vessels, called angiogenesis, is also an essential step in tumor growth in order for the tumor to get the blood supply it needs to expand. Variation in these genes may be predictive of any form of heart disease, numerous blood clotting disorders, stroke, hypertension and predisposition to tumor formation and metastasis. In particular, these variants may be predictive of the response to various antihypertensive drugs and chemotherapeutic and anti-tumor agents.

### **Apoptosis-related proteins**

Active cell suicide (apoptosis) is induced by events such as growth factor withdrawal and toxins. It is controlled by regulators, which have either an inhibitory effect on programmed cell death (anti-apoptotic) or block the protective effect of inhibitors (pro-apoptotic). Many viruses have found a way of countering defensive apoptosis by encoding their own anti-apoptosis genes preventing their target-cells from dying too soon. Variants of apoptosis related genes may be useful in formulation of antiaging drugs.

### **Cadherin, Cyclin, Polymerase, Oncogenes, Histones, Kinases**

Members of the cell division/cell cycle pathways such as cyclins, many transcription factors and kinases, DNA polymerases, histones, helicases and other oncogenes play a critical role in carcinogenesis where the uncontrolled proliferation of cells leads to tumor formation and eventually metastasis. Variation in these genes may be predictive of predisposition to any form of cancer, from increased risk of tumor formation to increased rate of metastasis. In particular, these variants may be predictive of the response to various chemotherapeutic and anti-tumor agents.

**Colony-stimulating factor-related proteins**

Granulocyte/macrophage colony-stimulating factors are cytokines that act in hematopoiesis by controlling the production, differentiation, and function of 2 related white cell populations of the blood, the granulocytes and the monocytes-macrophages.

**5 Complement-related proteins**

Complement proteins are immune associated cytotoxic agents, acting in a chain reaction to exterminate target cells that were opsonized (primed) with antibodies, by forming a membrane attack complex (MAC). The mechanism of killing is by opening pores in the target cell membrane. Variations in 20 complement genes or their inhibitors are 10 associated with many autoimmune disorders. Modified serum levels of complement products cause edemas of various tissues, lupus (SLE), vasculitis, glomerulonephritis, renal failure, hemolytic anemia, thrombocytopenia, and arthritis. They interfere with mechanisms of ADCC (antibody dependent cell cytotoxicity), severely impair immune competence and reduce phagocytic ability. Variants of complement genes may also be indicative of type I 15 diabetes mellitus, meningitis neurological disorders such as Nemaline myopathy, Neonatal hypotonia, muscular disorders such as congenital myopathy and other diseases.

**Cytochrome**

The respiratory chain is a key biochemical pathway which is essential to all aerobic cells. There are five different cytochromes involved in the chain. These are heme bound 20 proteins which serve as electron carriers. Modifications in these genes may be predictive of ataxia areflexia, dementia and myopathic and neuropathic changes in muscles. Also, association with various types of solid tumors.

**Kinesins**

Kinesins are tubulin molecular motors that function to transport organelles within 25 cells and to move chromosomes along microtubules during cell division. Modifications of these genes may be indicative of neurological disorders such as Pick disease of the brain, tuberous sclerosis.

## Cytokines, Interferon, Interleukin

Members of the cytokine families are known for their potent ability to stimulate cell growth and division even at low concentrations. Cytokines such as erythropoietin are cell-specific in their growth stimulation; erythropoietin is useful for the stimulation of the proliferation of erythroblasts. Variants in cytokines may be predictive for a wide variety of diseases, including cancer predisposition.

### G-protein coupled receptors

G-protein coupled receptors (also called R7G) are an extensive group of hormones, neurotransmitters, odorants and light receptors which transduce extracellular signals by interaction with guanine nucleotide-binding (G) proteins. Alterations in genes coding for G-coupled proteins may be involved in and indicative of a vast number of physiological conditions. These include blood pressure regulation, renal dysfunctions, male infertility, dopamine associated cognitive, emotional, and endocrine functions, hypercalcemia, chondrodysplasia and osteoporosis, pseudohypoparathyroidism, growth retardation and dwarfism.

### Thioesterases

Eukaryotic thiol proteases are a family of proteolytic enzymes which contain an active site cysteine. Catalysis proceeds through a thioester intermediate and is facilitated by a nearby histidine side chain; an asparagine completes the essential catalytic triad. Variants of thioester associated genes may be predictive of neuronal disorders and mental illnesses such as Ceroid Lipofuscinoses, Neuronal 1, Infantile, Santavuori disease and more.

### Breakdown Classifications of SNPs

The following list describes the numerical breakdown by molecule type of the SNPs described in Table 1. The key to these molecule types is as follows.

25

|    |                   |      |
|----|-------------------|------|
|    | TPase_associated: | 864  |
|    | Guanylyl:         | 3    |
|    | MHC:              | 1077 |
|    | amylase:          | 44   |
| 30 | amylaseinhib:     | 1    |
|    | amyloid:          | 96   |
|    | apoptosis:        | 91   |

|    |                     |      |
|----|---------------------|------|
|    | apoptosisinhib:     | 29   |
|    | apoptosisrecep:     | 14   |
|    | biotindep:          | 29   |
|    | cadhenn:            | 415  |
| 5  | calcium_channel:    | 85   |
|    | carboxylase:        | 4    |
|    | cathepsin:          | 336  |
|    | cathepsininhib:     | 41   |
|    | chloride_channel:   | 90   |
| 10 | collagen:           | 1542 |
|    | complement:         | 222  |
|    | complementinhib:    | 21   |
|    | complementrecept:   | 10   |
|    | csf:                | 31   |
| 15 | csf recept:         | 37   |
|    | cyclin:             | 65   |
|    | cyto45O:            | 136  |
|    | cytochrome:         | 659  |
|    | deaminase:          | 44   |
| 20 | dehydrogenase:      | 1235 |
|    | desaturase:         | 9    |
|    | dna_rna_bind:       | 1309 |
|    | dna_rna_bind_inhib: | 16   |
|    | dynein:             | 108  |
| 25 | elastase:           | 134  |
|    | elastaseinhib:      | 6    |
|    | eph:                | 487  |
|    | esterase:           | 258  |
|    | esteraseinhib:      | 3    |
| 30 | fgf:                | 34   |
|    | fgf receptor:       | 12   |
|    | gaba:               | 45   |
|    | glucoamylase:       | 106  |
|    | glucuronidase:      | 14   |
| 35 | glycoprotein:       | 3176 |
|    | helicase:           | 333  |
|    | histone:            | 272  |
|    | homeobox:           | 431  |
|    | hydrolase:          | 187  |
| 40 | hydroxysteroid:     | 84   |
|    | hypoxanthine:       | 4    |
|    | immunoglob:         | 1106 |
|    | immunoglob_recept:  | 19   |
|    | interferon:         | 322  |
| 45 | interleukin:        | 88   |
|    | interleukinrecept:  | 126  |
|    | isomerase:          | 404  |
|    | isomeraseinhibitor: | 45   |
|    | isomerasereceptor:  | 4    |
| 50 | kinase:             | 1684 |

|    |                    |       |
|----|--------------------|-------|
|    | kinase inhibitor:  | 187   |
|    | kinase receptor:   | 233   |
|    | kinesin:           | 86    |
|    | laminin:           | 196   |
| 5  | lipase:            | 63    |
|    | metallothionein:   | 62    |
|    | misc_channel:      | 215   |
|    | ngf:               | 30    |
|    | nucl_recpt:        | 339   |
| 10 | nuclease:          | 298   |
|    | oncogene:          | 783   |
|    | oxidase:           | 128   |
|    | oxygenase:         | 14    |
|    | peptidase:         | 150   |
| 15 | peroxidase:        | 115   |
|    | phosphatase:       | 668   |
|    | phosphataseinhib:  | 71    |
|    | phosphorylase:     | 84    |
|    | polymerase:        | 489   |
| 20 | potassium_channel: | 43    |
|    | prostaglandin:     | 55    |
|    | protease:          | 954   |
|    | proteaseinhib:     | 271   |
|    | reductase:         | 243   |
| 25 | ribosomal prot:    | 1040  |
|    | struct:            | 3128  |
|    | sulfotransferase:  | 42    |
|    | synthase:          | 893   |
|    | tgf:               | 117   |
| 30 | tgfreceptor:       | 41    |
|    | thioesterase:      | 3     |
|    | thiolase:          | 38    |
|    | tm7:               | 453   |
|    | tnf:               | 151   |
| 35 | tnfreceptor:       | 36    |
|    | traffic:           | 22    |
|    | transcriptfactor:  | 1139  |
|    | transferase:       | 291   |
|    | transport:         | 900   |
| 40 | tubulin:           | 334   |
|    | ubiquitin:         | 229   |
|    | water_channel:     | 18    |
|    | unclassified:      | 10567 |

The key to the molecule type is as follows:

|    | <b>Abbrev:</b>  | <b>Title:</b>  |
|----|---|--|
| 5  | amylase<br>amylaseinhib<br>amyloid<br>apoptosis<br>apoptosisinhib               | amylase protein<br>amylase inhibitor<br>amyloid protein<br>apoptosis associated protein<br>apoptosis inhibitors                            |
| 10 | apoptosisrecep<br>ATPase_associated<br>biotindep<br>cadherin<br>calcium_channel | apoptosis receptors<br>ATPase associated protein<br>biotin dependent enzyme/protein<br>cadherin protein<br>calcium channel protein         |
| 15 | carboxylase<br>cathepsin<br>cathepsininhib<br>chloride_channel                  | carboxylase protein<br>cathepsin/carboxypeptidases<br>cathepsin/carboxypeptidase inhibitor<br>chloride channel protein                     |
| 20 | collagen<br>complement<br>complementrecept<br>complementinhib                   | collagen<br>complement protein<br>complement receptor protein<br>complement inhibitor  |
| 25 | csf<br>csfrecept<br>cyclin<br>cyto450<br>cytochrome                             | colony stimulating factor<br>colony stimulating factor receptor<br>cyclin protein<br>cytochrome p450 protein<br>cytochrome related protein |
| 30 | deaminase<br>dehydrogenase<br>desaturase<br>dna_rna_bind                        | deaminase<br>dehydrogenase<br>desaturase<br>DNA/RNA binding protein/factor   |
|    | dna_rna_inhib   | DNA/RNA binding protein/factor inhibitor   |
| 35 | dynein<br>elastase<br>elastaseinhib<br>eph<br>esterase<br>esteraseinhib         | dynein<br>elastase<br>elastase inhibitor<br>EPH family of tyrosine kinases<br>esterase<br>esterase inhibitor                               |
| 40 | fgf<br>fgfreceptor<br>gaba<br>glucoamylase<br>glucoronidase                     | fibroblast growth factor<br>fibroblast growth factor receptor<br>GABA receptor<br>glucoamylase<br>glucuronidase                            |
| 45 | glycoprotein<br>Guanylyl<br>helicase<br>histone<br>HOM                          | glycoprotein<br>guanylylate cyclase<br>helicase<br>histone<br>homologous   |

|    |                    |   |
|----|--------------------|---|
|    | homeobox           | homeobox protein  |
|    | hydrolase          | hydrolase   |
|    | hydroxysteroid     | hydroxysteroid associated protein   |
|    | hypoxanthine       | hypoxanthine associated protein   |
| 5  | immunoglob         | immunoglobulin  |
|    | immunoglobrecept   | immunoglobulin receptor   |
|    | interferon         | interferon  |
|    | interleukin        | interleukin   |
|    | interleukinrecept  | interleukin receptor  |
| 10 | isomerase          | isomerase   |
|    | isomeraseinhibitor | isomerase inhibitor   |
|    | isomeraseseceptor  | isomerase receptor  |
|    | kinase             | kinase  |
|    | kinaseinhibitor    | kinase inhibitor  |
| 15 | kinasereceptor     | kinase receptor   |
|    | kinesin            | kinesin   |
|    | laminin            | laminin associated protein  |
|    | lipase             | lipase  |
|    | metallothionein    | metallothionein   |
| 20 | MHC                | major histocompatibility complex  |
|    | misc_channel       | miscellaneous channel   |
|    | ngf                | nerve growth factor   |
|    | nuci_recpt         | nuclear receptor  |
|    | nuclease           | nuclease  |
| 25 | oncogene           | oncogene associated protein   |
|    | oxidase            | oxidase   |
|    | oxygenase          | oxygenase   |
|    | peptidase          | peptidase   |
|    | peroxidase         | peroxidase  |
| 30 | phosphatase        | phosphatase   |
|    | phosphataseinhib   | phosphatase inhibitor   |
|    | phosphorylase      | phosphorylase   |
|    | PIR                | PIR DATABASE (release 56, 29-OCT-1998)  |
| 35 | polymerase         | polymerase  |
|    | potassium_channel  | potassium channel protein   |
|    | prostaglandin      | prostaglandin   |
|    | protease           | protease  |
|    | proteaseinhib      | protease inhibitor  |
| 40 | reductase          | reductase   |
|    | ribosomalprot      | ribosomal associated protein  |
|    | RTR                | EMBLDATABASE translated entries<br>not to be incorporated into SWISS-PROT (20-JUL-1998) |
| 45 | SIM                | similar   |
|    | SPTR               | EMBL DATABASE translated entries to<br>be incorporated into SWISS-PROT (20-JUL-1998)    |
|    | struct             | structural associated protein   |
| 50 | sulfotransferase   | sulfotransferase  |

|    |                  |   |
|----|------------------|---|
|    | SWP              | SWISS-PROT DATABASE (release 18-OCT-1998)                               |
|    | SWPN             | SWISS-PROT Update (release 11-NOV-98)                                   |
| 5  | synthase         | synthase  |
|    | tgf              | transforming growth factor  |
|    | tgfreceptor      | transforming growth factor receptor                                     |
|    | thioesterase     | thioesterase  |
|    | thiolase         | thiolase  |
| 10 | tm7              | seven transmembrane domain G-protein coupled receptor                   |
|    | tnf              | necrosis factor receptor  |
|    | traffic          | tumor necrosis factor   |
|    | tnfreceptor      | tumor trafficking associated protein                                    |
| 15 | TRN              | EMBL DATABASE translated entries update (20-JUL-1998)                   |
|    | transcriptfactor | transcription factor  |
|    | transferase      | transferase   |
|    | transport        | transport protein   |
| 20 | tubulin          | tubulin   |
|    | ubiquitin        | ubiquitin   |
|    | unclassified     | Protein not categorized into one of the aforementioned protein families |
|    | water channel    | water channel protein   |
| 25 |                  |   |

**Table 1**

A compilation of polymorphisms is listed in Table 1. Table 1 includes thirteen columns that provide descriptive information for each cSNP, each of which occupies one row in the Table. The column headings, and an explanation for each, are given below.

30        The first column of the table lists the names assigned to the fragments in which the polymorphisms occur. The fragments are all human genomic fragments. The sequence of one allelic form of each of the fragments (arbitrarily referred to as the prototypical or reference form) has been previously published. These sequences are listed at <http://www-genome.wi.mit.edu/> (all STS's sequence tag sites)); <http://shgc.stanford.edu> (Stanford STS's); and <http://www.tigr.org/> (TIGR STS's). The web sites also list primers for amplification of the fragments, and the genomic location of the fragments. Some fragments are expressed sequence tags, and some are random genomic fragments. All information in the web sites concerning the fragments listed in the table is incorporated by reference in its entirety for all purposes.

The second column lists the position in the fragment in which a polymorphic site has been found. Positions are numbered consecutively with the first base of the fragment sequence listed as in one of the above databases being assigned the number one. The third column lists the base occupying the polymorphic site in the sequence in the data base. This 5 base is arbitrarily designated the reference or prototypical form, but it is not necessarily the most frequently occurring form. The fourth column in the table lists the alternative base(s) at the polymorphic site. The fifth column of the table lists a 5' (upstream or forward) primer that hybridizes with the 5' end of the DNA sequence to be amplified. The sixth column of the 10 table lists a 3' (downstream or reverse) primer that hybridizes with the complement of the 3' end of the sequence to be amplified. The seventh column of the table lists a number of bases 15 of sequence on either side of the polymorphic site in each fragment. The indicated sequences can either be DNA or RNA. In the latter, the T's shown in the table are replaced by U's. The base occupying the polymorphic site is indicated in EUTAC-IUB ambiguity code.

“SEQ ID” provides the cross-references to the two nucleotide SEQ ID NOS: for the 15 cognate pair, which are numbered consecutively, and, as explained below, amino acid SEQ ID NOS: as well, in the Sequence Listing of the application.

Each sequence entry in the Sequence Listing also includes a cross-reference to the CuraGen sequence ID, under the label “Accession number”. The first pair of SEQ ID NOS: given in the first column of each row of the Table is the SEQ ID NO: identifying the nucleic 20 acid sequence for the polymorphism. If a polymorphism carries an entry for the amino acid portion of the row, a third SEQ ID NO: appears in parentheses in the column “Amino acid before” (see below) for the reference amino acid sequence, and a fourth SEQ ID NO: appears in parentheses in the column “Amino acid after” (see below) for the polymorphic amino acid sequence . The latter SEQ ID NOS: refer to amino acid sequences giving the cognate 25 reference and polymorphic amino acid sequences that are the translation of the nucleotide polymorphism. If a polymorphism carries no entry for the protein portion of the row, only one pair SEQ ID NOS: is provided, in the first column.

“CuraGen sequence ID” provides CuraGen Corporation’s accession number.

“Base pos. of SNP” gives the numerical position of the nucleotide in the nucleic acid 30 at which the cSNP is found, as identified in this invention.

“Polymorphic sequence” provides a 51-base sequence with the polymorphic site at the 26<sup>th</sup> base in the sequence, as well as 25 bases from the reference sequence on the 5’ side and the 3’ side of the polymorphic site. The designation at the polymorphic site is enclosed in square brackets, and provides first, the reference nucleotide; second, a “slash (/”); and  
5 third, the polymorphic nucleotide. In certain cases the polymorphism is an insertion or a deletion. In that case, the position that is “unfilled” (i.e., the reference or the polymorphic position) is indicated by the word “gap”.

“Base before” provides the nucleotide present in the reference sequence at the position at which the polymorphism is found.

10 “Base after” provides the altered nucleotide at the position of the polymorphism.

“Amino acid before” provides the amino acid in the reference protein, if the polymorphism occurs in a coding region. This column also includes the SEQ ID NO: in parentheses for the translated reference amino acid sequence if the polymorphism occurs in a coding region.

15 “Amino acid after” provides the amino acid in the polymorphic protein, if the polymorphism occurs in a coding region. This column also includes the SEQ ID NO in parentheses for the translated polymorphic amino acid sequence if the polymorphism occurs in a coding region.

“Type of change” provides information on the nature of the polymorphism.

20 “SILENT-NONCODING” is used if the polymorphism occurs in a noncoding region of a nucleic acid. “SILENT-CODING” is used if the polymorphism occurs in a coding region of a nucleic acid of a nucleic acid and results in no change of amino acid in the translated polymorphic protein. “CONSERVATIVE” is used if the polymorphism occurs in a coding region of a nucleic acid and provides a change in which the altered amino acid falls in the  
25 same class as the reference amino acid. The classes are: 1) Aliphatic: Gly, Ala, Val, Leu, Ile; 2) Aromatic: Phe, Tyr, Trp; 3) Sulfur-containing: Cys, Met; 4) Aliphatic OH: Ser, Thr; 5) Basic: Lys, Arg, His; 6) Acidic: Asp, Glu, Asn, Gln; 7) Pro falls in none of the other classes; and 8) End defines a termination codon.

“NONCONSERVATIVE” is used if the polymorphism occurs in a coding region of a nucleic acid and provides a change in which the altered amino acid falls in a different class than the reference amino acid.

“FRAMESHIFT” relates to an insertion or a deletion. If the frameshift occurs in a 5 coding region, the Table provides the translation of the frameshifted codons 3' to the polymorphic site.

“Protein classification of CuraGen gene” provides a generic class into which the protein is classified. Multiple classes of proteins were identified as listed above in the discussion of Table 1.

10 “Name of protein identified following a BLASTX analysis of the CuraGen sequence” provides the database reference for the protein found to resemble the novel reference-polymorphism cognate pair most closely.

15 “Similarity (pvalue) following a BLASTX analysis” provides the pvalue, a statistical measure from the BLASTX analysis that the polymorphic sequence is similar to, and therefore an allele of, the reference, or wild-type, sequence. In the present application, a cutoff of pvalue > 1 x 10<sup>-50</sup> (entered, for example, as 1.0E-50 in the Table) is used to establish that the reference-polymorphic cognate pairs are novel. A pvalue < 1 x 10<sup>-50</sup> defines proteins considered to be already known.

20 “Map location” provides any information available at the time of filing related to localization of a gene on a chromosome.

The polymorphisms are arranged in Table 1 in the following order:

SEQ ID NOs: 1-422 are nucleotide sequences for SNPs that are silent.

SEQ ID NOs: 423-480 are nucleotide sequences for SNPs that lead to conservative amino acid changes.

25 SEQ ID NOs: 481-619 are nucleotide sequences for SNPs that lead to nonconservative amino acid changes.

SEQ ID NOs: 620-651 are nucleotide sequences for SNPs that involve a gap. With respect to the reference or wild-type sequence at the position of the polymorphism, the allelic

cSNP introduces an additional nucleotide (an insertion) or deletes a nucleotide (a deletion). An SNP that involves a gap generates a frame shift.

Also presented in the sequence listing filed herewith are predicted amino acid sequences encoded by the polymorphic sequences shown in Table 1.

5 SEQ ID NOs: 652-709 are the amino acid sequences centered at the polymorphic amino acid residue for the protein products provided by SNPs that lead to conservative amino acid changes. 7 or 8 amino acids on either side of the polymorphic site are shown. The order in which these sequences appear mirrors the order of presentation of the cognate nucleotide sequences, and is set forth in the Table.

10 SEQ ID NOs: 710-848 are the amino acid sequences centered at the polymorphic amino acid residue for the protein products provided by SNPs that lead to nonconservative amino acid changes. 7 or 8 amino acids on either side of the polymorphic site are shown. The order in which these sequences appear mirrors the order of presentation of the cognate nucleotide sequences, and is set forth in the Table.

15 SEQ ID NOs: 849-880 are the amino acid sequences centered at the polymorphic amino acid residue for the protein products provided by SNPs that lead to frameshift-induced amino acid changes. 7 or 8 amino acids on either side of the polymorphic site are shown. The order in which these sequences appear mirrors the order of presentation of the cognate nucleotide sequences, and is set forth in the Table.

20 Provided herein are compositions which include, or are capable of detecting, nucleic acid sequences having these polymorphisms, as well as methods of using nucleic acids.

### **Identification of Individuals Carrying SNPs**

Individuals carrying polymorphic alleles of the invention may be detected at either the DNA, the RNA, or the protein level using a variety of techniques that are well known in the art. Strategies for identification and detection are described in *e.g.*, EP 730,663, EP 717,113, and PCT US97/02102. The present methods usually employ pre-characterized polymorphisms. That is, the genotyping location and nature of polymorphic forms present at a site have already been determined. The availability of this information allows sets of probes to be designed for specific identification of the known polymorphic forms.

Many of the methods described below require amplification of DNA from target samples. This can be accomplished by e.g., PCR. (1989), B. for detecting polymorphisms. See generally PCR Technology: Principles and Applications for DNA Amplification (ed. H.A. Erlich, Freeman Press, NY, NY, 1992); PCR Protocols: A Guide to Methods and Applications (eds. Innis, et al., Academic Press, San Diego, CA, 1990); Mattila et al., Nucleic Acids Res. 19, 4967 (1991); Eckert et al., PCR Methods and Applications 1, 17 (1991); PCR (eds. McPherson et al., IRL Press, Oxford); and U.S. Patent 4,683,202.

The phrase "recombinant protein" or "recombinantly produced protein" refers to a peptide or protein produced using non-native cells that do not have an endogenous copy of DNA able to express the protein. In particular, as used herein, a recombinantly produced protein relates to the gene product of a polymorphic allele, i.e., a "polymorphic protein" containing an altered amino acid at the site of translation of the nucleotide polymorphism. The cells produce the protein because they have been genetically altered by the introduction of the appropriate nucleic acid sequence. The recombinant protein will not be found in association with proteins and other subcellular components normally associated with the cells producing the protein. The terms "protein" and "polypeptide" are used interchangeably herein.

The phrase "substantially purified" or "isolated" when referring to a nucleic acid, peptide or protein, means that the chemical composition is in a milieu containing fewer, or preferably, essentially none, of other cellular components with which it is naturally associated. Thus, the phrase "isolated" or "substantially pure" refers to nucleic acid preparations that lack at least one protein or nucleic acid normally associated with the nucleic acid in a host cell. It is preferably in a homogeneous state although it can be in either a dry or aqueous solution. Purity and homogeneity are typically determined using analytical chemistry techniques such as gel electrophoresis or high performance liquid chromatography. Generally, a substantially purified or isolated nucleic acid or protein will comprise more than 80% of all macromolecular species present in the preparation. Preferably, the nucleic acid or protein is purified to represent greater than 90% of all macromolecular species present. More preferably the nucleic acid or protein is purified to greater than 95%, and most preferably the nucleic acid or protein is purified to essential homogeneity, wherein other macromolecular species are not detected by conventional analytical procedures.

The genomic DNA used for the diagnosis may be obtained from any nucleated cells of the body, such as those present in peripheral blood, urine, saliva, buccal samples, surgical specimen, and autopsy specimens. The DNA may be used directly or may be amplified enzymatically in vitro through use of PCR (Saiki et al. Science 239:487-491 (1988)) or other in vitro amplification methods such as the ligase chain reaction (LCR) (Wu and Wallace Genomics 4:560-569 (1989)), strand displacement amplification (SDA) (Walker et al. Proc. Natl. Acad. Sci. U.S.A. 89:392-396 (1992)), self-sustained sequence replication (3SR) (Fahy et al. PCR Methods P&J& 1:25-33 (1992)), prior to mutation analysis.

The method for preparing nucleic acids in a form that is suitable for mutation detection is well known in the art. A "nucleic acid" is a deoxyribonucleotide or ribonucleotide polymer in either single-or double-stranded form, including known analogs of natural nucleotides unless otherwise indicated. The term "nucleic acids", as used herein, refers to either DNA or RNA. "Nucleic acid sequence" or "polynucleotide sequence" refers to a single-stranded sequence of deoxyribonucleotide or ribonucleotide bases read from the 5' end to the 3' end. The direction of 5' to 3' addition of nascent RNA transcripts is referred to as the transcription direction; sequence regions on the DNA strand having the same sequence as the RNA and which are beyond the 5' end of the RNA transcript in the 5' direction are referred to as "upstream sequences"; sequence regions on the DNA strand having the same sequence as the RNA and which are beyond the 3' end of the RNA transcript in the 3' direction are referred to as "downstream sequences". The term includes both self-replicating plasmids, infectious polymers of DNA or RNA and nonfunctional DNA or RNA. The complement of any nucleic acid sequence of the invention is understood to be included in the definition of that sequence. "Nucleic acid probes" may be DNA or RNA fragments.

The detection of polymorphisms in specific DNA sequences, can be accomplished by a variety of methods including, but not limited to, restriction-fragment-length-polymorphism detection based on allele-specific restriction-endonuclease cleavage (Kan and Dozy Lancet ii:910-912 (1978)), hybridization with allele-specific oligonucleotide probes (Wallace et al. Nucl. Acids Res. 6:3543-3557 (1978)), including immobilized oligonucleotides (Saiki et al. Proc. Natl. Acad. Sci. USA, 86:6230-6234 (1989)) or oligonucleotide arrays (Maskos and Southern Nucl. Acids Res. 21:2269-2270 (1993)), allele-specific PCR (Newton et al. Nucl. Acids Res. 17:2503-2516 (1989)), mismatch-repair detection (MRD) (Faham and Cox Genome Res. 5:474-482 (1995)), binding of MutS protein (Wagner et al. Nucl. Acids Res. 23:3944-3948 (1995)), denaturing-gradient gel electrophoresis (DGGE) (Fisher and Lerman et

al. *Proc. Natl. Acad. Sci. U.S.A.* 80:1579-1583 (1983)), single-strand-conformation-polymorphism detection (Orita et al. *Genomics* 5:874-879 (1983)), RNAase cleavage at mismatched base-pairs (Myers et al. *Science* 230:1242 (1985)), chemical (Cotton et al. *Proc. Natl. Acad. Sci. U.S.A.* 85:4397-4401 (1988)) or enzymatic (Youil et al. *Proc. Natl. Acad. Sci. U.S.A.* 92:87-91 (1995)) cleavage of heteroduplex DNA, methods based on allele specific primer extension (Syvanen et al. *Genomics* 8:684-692 (1990)), genetic bit analysis (GBA) (Nikiforov et al. *&&I Acids* 22:4167-4175 (1994)), the oligonucleotide-ligation assay (OLA) (Landegren et al. *Science* 241:1077 (1988)), the allele-specific ligation chain reaction (LCR) (Barrany *Proc. Natl. Acad. Sci. U.S.A.* 88:189-193 (1991)), gap-LCR (Abravaya et al. *Nucl. Acids Res.* 23:675-682 (1995)), radioactive and/or fluorescent DNA sequencing using standard procedures well known in the art, and peptide nucleic acid (PNA) assays (Orum et al., *Nucl. Acids Res.* 21:5332-5356 (1993); Thiede et al., *Nucl. Acids Res.* 24:983-984 (1996)).

“Specific hybridization” or “selective hybridization” refers to the binding, or duplexing, of a nucleic acid molecule only to a second particular nucleotide sequence to which the nucleic acid is complementary, under suitably stringent conditions when that sequence is present in a complex mixture (e.g., total cellular DNA or RNA). “Stringent conditions” are conditions under which a probe will hybridize to its target subsequence, but to no other sequences. Stringent conditions are sequence-dependent and are different in different circumstances. Longer sequences hybridize specifically at higher temperatures than shorter ones. Generally, stringent conditions are selected such that the temperature is about 5°C lower than the thermal melting point (Tm) for the specific sequence to which hybridization is intended to occur at a defined ionic strength and pH. The Tm is the temperature (under defined ionic strength, pH, and nucleic acid concentration) at which 50% of the target sequence hybridizes to the complementary probe at equilibrium. Typically, stringent conditions include a salt concentration of at least about 0.01 to about 1.0 M Na ion concentration (or other salts), at pH 7.0 to 8.3. The temperature is at least about 30°C for short probes (e.g., 10 to 50 nucleotides). Stringent conditions can also be achieved with the addition of destabilizing agents such as formamide. For example, conditions of 5X SSPE (750 mM NaCl, 50 mM NaPhosphate, 5 mM EDTA, pH 7.4) and a temperature of 25-30°C are suitable for allele-specific probe hybridizations.

“Complementary” or “target” nucleic acid sequences refer to those nucleic acid sequences which selectively hybridize to a nucleic acid probe. Proper annealing conditions

depend, for example, upon a probe's length, base composition, and the number of mismatches and their position on the probe, and must often be determined empirically. For discussions of nucleic acid probe design and annealing conditions, see, for example, Sambrook et al., or Current Protocols in Molecular Biology, F. Ausubel *et al.*, ed., Greene Publishing and Wiley-Interscience, New York (1987).

A perfectly matched probe has a sequence perfectly complementary to a particular target sequence. The test probe is typically perfectly complementary to a portion of the target sequence. A "polymorphic" marker or site is the locus at which a sequence difference occurs with respect to a reference sequence. Polymorphic markers include restriction fragment length polymorphisms, variable number of tandem repeats (VNTR's), hypervariable regions, minisatellites, dinucleotide repeats, trinucleotide repeats, tetranucleotide repeats, simple sequence repeats, and insertion elements such as Alu. The reference allelic form may be, for example, the most abundant form in a population, or the first allelic form to be identified, and other allelic forms are designated as alternative, variant or polymorphic alleles. The allelic form occurring most frequently in a selected population is sometimes referred to as the "wild type" form, and herein may also be referred to as the "reference" form. Diploid organisms may be homozygous or heterozygous for allelic forms. A diallelic polymorphism has two distinguishable forms (i.e., base sequences), and a triallelic polymorphism has three such forms.

As use herein an "oligonucleotide" is a single-stranded nucleic acid ranging in length from 2 to about 60 bases. Oligonucleotides are often synthetic but can also be produced from naturally occurring polynucleotides. A probe is an oligonucleotide capable of binding to a target nucleic acid of complementary sequence through one or more types of chemical bonds, usually through complementary base pairing via hydrogen bond formation. Oligonucleotides probes are often between 5 and 60 bases, and, in specific embodiments, may be between 10-20, or 15-30 bases long. An oligonucleotide probe may include natural (i.e. A, G, C, or T) or modified bases (7-deazaguanosine, inosine, etc.). In addition, the bases in an oligonucleotide probe may be joined by a linkage other than a phosphodiester bond, such as a phosphoramidite linkage or a phosphorothioate linkage, or they may be peptide nucleic acids in which the constituent bases are joined by peptide bonds rather than by phosphodiester bonds, so long as it does not interfere with hybridization. Examples of an oligonucleotide are shown in Table 1. Oligonucleotides can be all of a nucleic acid segment as represented in column 4 of Table 1; a nucleic acid sequence which comprises a nucleic acid segment

represented in column 4 of Table 1 and additional nucleic acids (present at either or both ends of a nucleic acid segment of column 4); or a portion (fragment) of a nucleic acid segment represented in column 4 of the table which includes a polymorphic site. Preferred polymorphic sites of the invention include segments of DNA or their complements, which 5 include any one of the polymorphic sites shown in the Table. The segments can be between 5 and 250 bases, and, in specific embodiments are between 5-10, 5-20, 10-20, 10-50, 20-50 or 10-100 bases. The polymorphic site can occur within any position of the segment. The segments can be from any of the allelic forms of the DNA shown in the Table.

As used herein, the term "primer" refers to a single-stranded oligonucleotide which 10 acts as a point of initiation of template-directed DNA synthesis under appropriate conditions (e.g., in the presence of four different nucleoside triphosphates and a polymerization agent, such as DNA polymerase, RNA polymerase or reverse transcriptase) in an appropriate buffer and at a suitable temperature. The appropriate length of a primer depends on the intended use of the primer, but typically ranges from 15 to 30 nucleotides. Short primer molecules 15 generally require cooler temperatures to form sufficiently stable hybrid complexes with the template. A primer need not be perfectly complementary to the exact sequence of the template, but should be sufficiently complementary to hybridize with it. The term "primer site" refers to the sequence of the target DNA to which a primer hybridizes. The term "primer pair" refers to a set of primers including a 5' (upstream) primer that hybridizes with 20 the 5' end of the DNA sequence to be amplified and a 3' (downstream) primer that hybridizes with the complement of the 3' end of the sequence to be amplified.

DNA fragments can be prepared, for example, by digesting plasmid DNA, or by use 25 of PCR. Oligonucleotides for use as primers or probes are chemically synthesized by methods known in the field of the chemical synthesis of polynucleotides, including by way of non-limiting example the phosphoramidite method described by Beaucage and Carruthers, Tetrahedron Lett 22:1859-1862 (1981) and the triester method provided by Matteucci, et al., J. Am. Chem. Soc., 103:3185 (1981) both incorporated herein by reference. These syntheses may employ an automated synthesizer, as described in Needham-VanDevanter, D.R., et al., Nucleic Acids Res. 12:61596168 (1984). Purification of oligonucleotides may 30 be carried out by either native acrylamide gel electrophoresis or by anion-exchange HPLC as described in Pearson, J.D. and Regnier, F.E., J. Chrom., 255:137-149 (1983). A double stranded fragment may then be obtained, if desired, by annealing appropriate complementary single strands together under suitable conditions or by synthesizing the complementary strand

using a DNA polymerase with an appropriate primer sequence. Where a specific sequence for a nucleic acid probe is given, it is understood that the complementary strand is also identified and included. The complementary strand will work equally well in situations where the target is a double-stranded nucleic acid.

5        The sequence of the synthetic oligonucleotide or of any nucleic acid fragment can be  
can be obtained using either the dideoxy chain termination method or the Maxam-Gilbert  
method (see Sambrook et al. Molecular Cloning - a Laboratory Manual (2nd Ed.), Vols. 1-  
3, Cold Spring Harbor Laboratory, Cold Spring Harbor, New York, (1989), which is  
incorporated herein by reference. This manual is hereinafter referred to as "Sambrook et al."  
10 ; Zyskind et al., (1988)). Recombinant DNA Laboratory Manual, (Acad. Press, New York).  
Oligonucleotides useful in diagnostic assays are typically at least 8 consecutive nucleotides in  
length, and may range upwards of 18 nucleotides in length to greater than 100 or more  
consecutive nucleotides.

15      Another aspect of the invention pertains to isolated antisense nucleic acid molecules  
that are hybridizable to or complementary to the nucleic acid molecule comprising the SNP-  
containing nucleotide sequences of the invention, or fragments, analogs or derivatives  
thereof. An "antisense" nucleic acid comprises a nucleotide sequence that is complementary  
to a "sense" nucleic acid encoding a protein, e.g., complementary to the coding strand of a  
double-stranded cDNA molecule or complementary to an mRNA sequence. In specific  
20 aspects, antisense nucleic acid molecules are provided that comprise a sequence  
complementary to at least about 10, about 25, about 50, or about 60 nucleotides or an entire  
SNP coding strand, or to only a portion thereof.

25      In one embodiment, an antisense nucleic acid molecule is antisense to a "coding  
region" of the coding strand of a polymorphic nucleotide sequence of the invention. The term  
"coding region" refers to the region of the nucleotide sequence comprising codons which are  
translated into amino acid. In another embodiment, the antisense nucleic acid molecule is  
antisense to a "noncoding region" of the coding strand of a nucleotide sequence of the  
invention. The term "noncoding region" refers to 5' and 3' sequences which flank the coding  
region that are not translated into amino acids (*i.e.*, also referred to as 5' and 3' untranslated  
30 regions).

Given the coding strand sequences disclosed herein, antisense nucleic acids of the invention can be designed according to the rules of Watson and Crick or Hoogsteen base pairing. For example, the antisense nucleic acid molecule can generally be complementary to the entire coding region of an mRNA, but more preferably as embodied herein, it is an oligonucleotide that is antisense to only a portion of the coding or noncoding region of the mRNA. An antisense oligonucleotide can range in length between about 5 and about 60 nucleotides, preferably between about 10 and about 45 nucleotides, more preferably between about 15 and 40 nucleotides, and still more preferably between about 15 and 30 in length. An antisense nucleic acid of the invention can be constructed using chemical synthesis or enzymatic ligation reactions using procedures known in the art. For example, an antisense nucleic acid (*e.g.*, an antisense oligonucleotide) can be chemically synthesized using naturally occurring nucleotides or variously modified nucleotides designed to increase the biological stability of the molecules or to increase the physical stability of the duplex formed between the antisense and sense nucleic acids, *e.g.*, phosphorothioate derivatives and acridine substituted nucleotides can be used.

Examples of modified nucleotides that can be used to generate the antisense nucleic acid include: 5-fluorouracil, 5-bromouracil, 5-chlorouracil, 5-iodouracil, hypoxanthine, xanthine, 4-acetylcytosine, 5-(carboxyhydroxymethyl) uracil, 5-carboxymethylaminomethyl-2-thiouridine, 5-carboxymethylaminomethyluracil, dihydrouracil, beta-D-galactosylqueosine, inosine, N6-isopentenyladenine, 1-methylguanine, 1-methylinosine, 2,2-dimethylguanine, 2-methyladenine, 2-methylguanine, 3-methylcytosine, 5-methylcytosine, N6-adenine, 7-methylguanine, 5-methylaminomethyluracil, 5-methoxyaminomethyl-2-thiouracil, beta-D-mannosylqueosine, 5'-methoxycarboxymethyluracil, 5-methoxyuracil, 2-methylthio-N6-isopentenyladenine, uracil-5-oxyacetic acid (v), wybutoxosine, pseudouracil, queosine, 2-thiocytosine, 5-methyl-2-thiouracil, 2-thiouracil, 4-thiouracil, 5-methyluracil, uracil-5-oxyacetic acid methylester, uracil-5-oxyacetic acid (v), 5-methyl-2-thiouracil, 3-(3-amino-3-N-2-carboxypropyl) uracil, (acp3)w, and 2,6-diaminopurine. Alternatively, the antisense nucleic acid can be produced biologically using an expression vector into which a nucleic acid has been subcloned in an antisense orientation (*i.e.*, RNA transcribed from the inserted nucleic acid will be of an antisense orientation to a target nucleic acid of interest, described further in the following subsection).

The antisense nucleic acid molecules of the invention are typically administered to a subject or generated *in situ* such that they hybridize with or bind to cellular mRNA and/or

genomic DNA encoding a polymorphic protein to thereby inhibit expression of the protein, e.g., by inhibiting transcription and/or translation. The hybridization can be by conventional nucleotide complementary to form a stable duplex, or, for example, in the case of an antisense nucleic acid molecule that binds to DNA duplexes, through specific interactions in the major groove of the double helix. An example of a route of administration of antisense nucleic acid molecules of the invention includes direct injection at a tissue site.

Alternatively, antisense nucleic acid molecules can be modified to target selected cells and then administered systemically. For example, for systemic administration, antisense molecules can be modified such that they specifically bind to receptors or antigens expressed on a selected cell surface, e.g., by linking the antisense nucleic acid molecules to peptides or antibodies that bind to cell surface receptors or antigens. The antisense nucleic acid molecules can also be delivered to cells using the vectors described herein. To achieve sufficient intracellular concentrations of antisense molecules, vector constructs in which the antisense nucleic acid molecule is placed under the control of a strong pol II or pol III promoter are preferred.

In yet another embodiment, the antisense nucleic acid molecule of the invention is an  $\alpha$ -anomeric nucleic acid molecule. An  $\alpha$ -anomeric nucleic acid molecule forms specific double-stranded hybrids with complementary RNA in which, contrary to the usual  $\beta$ -units, the strands run parallel to each other (Gaultier *et al.* (1987) *Nucleic Acids Res* 15: 6625-6641).

The antisense nucleic acid molecule can also comprise a 2'-o-methylribonucleotide (Inoue *et al.* (1987) *Nucleic Acids Res* 15: 6131-6148) or a chimeric RNA -DNA analogue (Inoue *et al.* (1987) *FEBS Lett* 215: 327-330).

The following terms are used to describe the sequence relationships between two or more nucleic acids or polynucleotides: "reference sequence", "comparison window", "sequence identity", "percentage of sequence identity", and "substantial identity". A "reference sequence" is a defined sequence used as a basis for a sequence comparison; a reference sequence may be a subset of a larger sequence, for example, as a segment of a full-length cDNA or gene sequence given in a sequence listing, or may comprise a complete cDNA or gene sequence. Optimal alignment of sequences for aligning a comparison window may, for example, be conducted by the local homology algorithm of Smith and Waterman *Adv. Appl. Math.* 2482 (1981), by the homology alignment algorithm of Needleman and Wunsch *J. Mol. Biol.* 48:443 (1970), by the search for similarity method of Pearson and

Lipman Proc. Natl. Acad. Sci. U.S.A. 852444 (1988), or by computerized implementations of these algorithms (for example, GAP, BESTFIT, FASTA, and TFASTA in the Wisconsin Genetics Software Package Release 7.0, Genetics Computer Group, 575 Science Dr., Madison, WI).

5 Techniques for nucleic acid manipulation of the nucleic acid sequences harboring the cSNP's of the invention, such as subcloning nucleic acid sequences encoding polypeptides into expression vectors, labeling probes, DNA hybridization, and the like, are described generally in Sambrook et al., The phrase "nucleic acid sequence encoding" refers to a nucleic acid which directs the expression of a specific protein, peptide or amino acid sequence. The  
10 nucleic acid sequences include both the DNA strand sequence that is transcribed into RNA and the RNA sequence that is translated into protein, peptide or amino acid sequence. The nucleic acid sequences include both the full length nucleic acid sequences disclosed herein as well as non-full length sequences derived from the full length protein. It being further understood that the sequence includes the degenerate codons of the native sequence or  
15 sequences which may be introduced to provide codon preference in a specific host cell. Consequently, the principles of probe selection and array design can readily be extended to analyze more complex polymorphisms (see EP 730,663). For example, to characterize a triallelic SNP polymorphism, three groups of probes can be designed tiled on the three polymorphic forms as described above. As a further example, to analyze a diallelic  
20 polymorphism involving a deletion of a nucleotide, one can tile a first group of probes based on the undeleted polymorphic form as the reference sequence and a second group of probes based on the deleted form as the reference sequence.

For assay of genomic DNA, virtually any biological convenient tissue samples include whole blood, semen, saliva, tears, urine, fecal material, sweat, buccal, skin and hair  
25 can be used. Genomic DNA is typically amplified before analysis. Amplification is usually effected by PCR using primers flanking a suitable fragment e.g., of 50-500 nucleotides containing the locus of the polymorphism to be analyzed. Target is usually labeled in the course of amplification. The amplification product can be RNA or DNA, single stranded or double stranded. If double stranded, the amplification product is typically denatured before  
30 application to an array. If genomic DNA is analyzed without amplification, it may be desirable to remove RNA from the sample before applying it to the array. Such can be accomplished by digestion with DNase-free RNase.

**DETECTION OF POLYMORPHISMS IN A NUCLEIC ACID SAMPLE**

The SNPs disclosed herein can be used to determine which forms of a characterized polymorphism are present in individuals under analysis.

The design and use of allele-specific probes for analyzing polymorphisms is described by e.g., Saiki et al., Nature 324, 163-166 (1986); Dattagupta, EP 235,726, Saiki, WO 89/11548. Allele-specific probes can be designed that hybridize to a segment of target DNA from one individual but do not hybridize to the corresponding segment from another individual due to the presence of different polymorphic forms in the respective segments from the two individuals. Hybridization conditions should be sufficiently stringent that there is a significant difference in hybridization intensity between alleles, and preferably an essentially binary response, whereby a probe hybridizes to only one of the alleles. Some probes are designed to hybridize to a segment of target DNA such that the polymorphic site aligns with a central position (e.g., in a 15-mer at the 7 position; in a 16-mer, at either the 7, 8 or 9 position) of the probe. This design of probe achieves good discrimination in hybridization between different allelic forms.

Allele-specific probes are often used in pairs, one member of a pair showing a perfect match to a reference form of a target sequence and the other member showing a perfect match to a variant form. Several pairs of probes can then be immobilized on the same support for simultaneous analysis of multiple polymorphisms within the same target sequence.

The polymorphisms can also be identified by hybridization to nucleic acid arrays, some examples of which are described in published PCT application WO 95/11995. WO 95/11995 also describes subarrays that are optimized for detection of a variant form of a precharacterized polymorphism. Such a subarray contains probes designed to be complementary to a second reference sequence, which is an allelic variant of the first reference sequence. The second group of probes is designed by the same principles, except that the probes exhibit complementarity to the second reference sequence. The inclusion of a second group (or further groups) can be particularly useful for analyzing short subsequences of the primary reference sequence in which multiple mutations are expected to occur within a short distance commensurate with the length of the probes (e.g., two or more mutations within 9 to 21 bases).

An allele-specific primer hybridizes to a site on target DNA overlapping a polymorphism and only primes amplification of an allelic form to which the primer exhibits perfect complementarity. See Gibbs, Nucleic Acid Res. 17 2427-2448 (1989). This primer is used in conjunction with a second primer which hybridizes at a distal site.

5      Amplification proceeds from the two-primers, resulting in a detectable product which indicates the particular allelic form is present. A control is usually performed with a second pair of primers, one of which shows a single base mismatch at the polymorphic site and the other of which exhibits perfect complementarity to a distal site. The single-base mismatch prevents amplification and no detectable product is formed. The method works best when  
10     the mismatch is included in the 3'-most position of the oligonucleotide aligned with the polymorphism because this position is most destabilizing to elongation from the primer (see, e.g., WO 93/22456).

Amplification products generated using the polymerase chain reaction can be analyzed by the use of denaturing gradient gel electrophoresis. Different alleles can be  
15     identified based on the different sequence-dependent melting properties and electrophoretic migration of DNA in solution. Erlich, ed., PCR Technology, Principles and Applications for DNA Amplification, (W.H. Freeman and Co New York, 1992, Chapter 7).

Alleles of target sequences can be differentiated using single-strand conformation polymorphism analysis, which identifies base differences by alteration in electrophoretic  
20     migration of single stranded PCR products, as described in Orita et al., Proc. Nat. Acad. Sci. 86, 2766-2770 (1989). Amplified PCR products can be generated and heated or otherwise denatured, to form single stranded amplification products. Single-stranded nucleic acids may refold or form secondary structures which are partially dependent on the base sequence. The different electrophoretic mobilities of single-stranded amplification  
25     products can be related to base-sequence differences between alleles of target sequences.

The genotype of an individual with respect to a pathology suspected of being caused by a genetic polymorphism may be assessed by association analysis. Phenotypic traits suitable for association analysis include diseases that have known but hitherto unmapped genetic components (e.g., agammaglobulinemia, diabetes insipidus, Lesch-Nyhan syndrome,  
30     muscular dystrophy, Wiskott-Aldrich syndrome, Fabry's disease, familial hypercholesterolemia, polycystic kidney disease, hereditary spherocytosis, von Willebrand's

disease, tuberous sclerosis, hereditary hemorrhagic telangiectasia, familial colonic polyposis, Ehlers-Danlos syndrome, osteogenesis imperfecta, and acute intermittent porphyria).

Phenotypic traits also include symptoms of, or susceptibility to, multifactorial diseases of which a component is or may be genetic, such as autoimmune diseases, 5 inflammation, cancer, system, diseases of the nervous and infection by pathogenic microorganisms. Some examples of autoimmune diseases include rheumatoid arthritis, multiple sclerosis, diabetes (insulin-dependent and non-independent), systemic lupus erythematosus and Graves disease. Some examples of cancers include cancers of the bladder, brain, breast, colon, esophagus, kidney, oral cavity, ovary, pancreas, prostate, skin, stomach, 10 leukemia, liver, lung, and uterus. Phenotypic traits also include characteristics such as longevity, appearance (e.g., baldness, obesity), strength, speed, endurance, fertility, and susceptibility or receptivity to particular drugs or therapeutic treatments.

Such correlations can be exploited in several ways. In the case of a strong correlation between a polymorphic form and a disease for which treatment is available, detection of the 15 polymorphic form set in a human or animal patient may justify immediate administration of treatment, or at least the institution of regular monitoring of the patient. Detection of a polymorphic form correlated with serious disease in a couple contemplating a family may also be valuable to the couple in their reproductive decisions. For example, the female partner might elect to undergo in vitro fertilization to avoid the possibility of transmitting 20 such a polymorphism from her husband to her offspring. In the case of a weaker, but still statistically significant correlation between a polymorphic set and human disease, immediate therapeutic intervention or monitoring may not be justified. Nevertheless, the patient can be motivated to begin simple life-style changes (e.g., diet, exercise) that can be accomplished at little cost to the patient but confer potential benefits in reducing the risk of conditions to 25 which the patient may have increased susceptibility by virtue of variant alleles. After determining polymorphic form(s) present in an individual at one or more polymorphic sites, this information can be used in a number of methods.

Determination of which polymorphic forms occupy a set of polymorphic sites in an individual identifies a set of polymorphic forms that distinguishes the individual. See 30 generally National Research Council, *The Evaluation of Forensic DNA Evidence* (Eds. Pollard et al., National Academy Press, DC, 1996). Since the polymorphic sites are within a 50,000 bp region in the human genome, the probability of recombination between these

polymorphic sites is low. That low probability means the haplotype (the set of all 10 polymorphic sites) set forth in this application should be inherited without change for at least several generations. The more sites that are analyzed the lower the probability that the set of polymorphic forms in one individual is the same as that in an unrelated individual.

5 Preferably, if multiple sites are analyzed, the sites are unlinked. Thus, polymorphisms of the invention are often used in conjunction with polymorphisms in distal genes. Preferred polymorphisms for use in forensics are diallelic because the population frequencies of two polymorphic forms can usually be determined with greater accuracy than those of multiple polymorphic forms at multi-allelic loci.

10 The capacity to identify a distinguishing or unique set of forensic markers in an individual is useful for forensic analysis. For example, one can determine whether a blood sample from a suspect matches a blood or other tissue sample from a crime scene by determining whether the set of polymorphic forms occupying selected polymorphic sites is the same in the suspect and the sample. If the set of polymorphic markers does not match  
15 between a suspect and a sample, it can be concluded (barring experimental error) that the suspect was not the source of the sample. If the set of markers does match, one can conclude that the DNA from the suspect is consistent with that found at the crime scene. If frequencies of the polymorphic forms at the loci tested have been determined (e.g., by analysis of a suitable population of individuals), one can perform a statistical analysis to determine the  
20 probability that a match of suspect and crime scene sample would occur by chance.

p(ID) is the probability that two random individuals have the same polymorphic or allelic form at a given polymorphic site. In diallelic loci, four genotypes are possible: AA, AB, BA, and BB. If alleles A and B occur in a haploid genome of the organism with frequencies x and y, the probability of each genotype in a diploid organism are (see WO  
25 95/12607):

$$\text{Homozygote: } p(AA)=x^2$$

$$\text{Homozygote: } p(BB)=y^2=(1-x)^2$$

$$\text{Single Heterozygote: } p(AB)=p(BA)=xy=x(1-x)$$

$$\text{Both Heterozygotes: } p(AB+BA)=2xy=2x(1-x)$$

The probability of identity at one locus (i.e. the probability that two individuals, picked at random from a population will have identical polymorphic forms at a given locus) is given by the equation:

$$p(ID) = (x^2)^2 + (2xy)^2 + (y^2)^2.$$

5 These calculations can be extended for any number of polymorphic forms at a given locus. For example, the probability of identity  $p(ID)$  for a 3-allele system where the alleles have the frequencies in the population of  $x$ ,  $y$  and  $z$ , respectively, is equal to the sum of the squares of the genotype frequencies:

$$p(ID) = x^4 + (2xy)^2 + (2yz)^2 + (2xz)^2 + z^4 + y^4$$

10 In a locus of  $n$  alleles, the appropriate binomial expansion is used to calculate  $p(ID)$  and  $p(exc)$ .

The cumulative probability of identity (cum  $p(ID)$ ) for each of multiple unlinked loci is determined by multiplying the probabilities provided by each locus:

$$cum\ p(ID) = p(ID_1)p(ID_2)p(ID_3)\dots p(ID_n)$$

15 The cumulative probability of non-identity for  $n$  loci (i.e. the probability that two random individuals will be different at 1 or more loci) is given by the equation:

$$cum\ p(nonID) = 1 - cum\ p(ID).$$

If several polymorphic loci are tested, the cumulative probability of non-identity for random individuals becomes very high (e.g., one billion to one). Such probabilities can be taken into account together with other evidence in determining the guilt or innocence of the suspect.

20 The object of paternity testing is usually to determine whether a male is the father of a child. In most cases, the mother of the child is known and thus, the mother's contribution to the child's genotype can be traced. Paternity testing investigates whether the part of the child's genotype not attributable to the mother is consistent with that of the putative father. Paternity testing can be performed by analyzing sets of polymorphisms in the putative father and the child.

If the set of polymorphisms in the child attributable to the father does not match the putative father, it can be concluded, barring experimental error, that the putative father is not the real father. If the set of polymorphisms in the child attributable to the father does match the set of polymorphisms of the putative father, a statistical calculation can be performed to determine the probability of coincidental match.

The probability of parentage exclusion (representing the probability that a random male will have a polymorphic form at a given polymorphic site that makes him incompatible as the father) is given by the equation (see WO 95/12607):

$$p(\text{exc})=xy(1-xy)$$

10 where x and y are the population frequencies of alleles A and B of a diallelic polymorphic site. (At a triallelic site  $p(\text{exc})=xy(1-xy)+yz(1-yz)+xz(1-xz)+3xyz(1-xyz))$ , where x, y and z are the respective population frequencies of alleles A, B and C). The probability of non-exclusion is:

$$p(\text{non-exc})=1-p(\text{exc})$$

15 The cumulative probability of non-exclusion (representing the value obtained when n loci are used) is thus:

$$\text{cum } p(\text{non-exc})=p(\text{non-exc}1)p(\text{non-exc}2)p(\text{non-exc}3)\dots p(\text{non-exc}n)$$

The cumulative probability of exclusion for n loci (representing the probability that a random male will be excluded) is:

20  $\text{cum } p(\text{exc})=1-\text{cum } p(\text{non-exc}).$

If several polymorphic loci are included in the analysis, the cumulative probability of exclusion of a random male is very high. This probability can be taken into account in assessing the liability of a putative father whose polymorphic marker set matches the child's polymorphic marker set attributable to his/her father.

25 The polymorphisms of the invention may contribute to the phenotype of an organism in different ways. Some polymorphisms occur within a protein coding sequence and contribute to phenotype by affecting protein structure. The effect may be neutral, beneficial or detrimental, or both beneficial and detrimental, depending on the circumstances. For

example, a heterozygous sickle cell mutation confers resistance to malaria, but a homozygous sickle cell mutation is usually lethal. Other polymorphisms occur in noncoding regions but may exert phenotypic effects indirectly via influence on replication, transcription, and translation. A single polymorphism may affect more than one phenotypic trait. Likewise, a 5 single phenotypic trait may be affected by polymorphisms in different genes. Further, some polymorphisms predispose an individual to a distinct mutation that is causally related to a certain phenotype.

Phenotypic traits include diseases that have known but hitherto unmapped genetic components. Phenotypic traits also include symptoms of, or susceptibility to, multifactorial 10 diseases of which a component is or may be genetic, such as autoimmune diseases, inflammation, cancer, diseases of the nervous system, and infection by pathogenic microorganisms. Some examples of autoimmune diseases include rheumatoid arthritis, multiple sclerosis, diabetes (insulin-dependent and non-independent), systemic lupus erythematosus and Graves disease. Some examples of cancers include cancers of the bladder, 15 brain, breast, colon, esophagus, kidney, leukemia, liver, lung, oral cavity, ovary, pancreas, prostate, skin, stomach and uterus. Phenotypic traits also include characteristics such as longevity, appearance (e.g., baldness, obesity), strength, speed, endurance, fertility, and susceptibility or receptivity to particular drugs or therapeutic treatments.

Correlation is performed for a population of individuals who have been tested for the 20 presence or absence of a phenotypic trait of interest and for polymorphic markers sets. To perform such analysis, the presence or absence of a set of polymorphisms (i.e. a polymorphic set) is determined for a set of the individuals, some of whom exhibit a particular trait, and some of which exhibit lack of the trait. The alleles of each polymorphism of the set are then reviewed to determine whether the presence or absence of a particular allele is associated 25 with the trait of interest. Correlation can be performed by standard statistical methods such as a  $\chi^2$ -squared test and statistically significant correlations between polymorphic form(s) and phenotypic characteristics are noted. For example, it might be found that the presence of allele A1 at polymorphism A correlates with heart disease. As a further example, it might be found that the combined presence of allele A1 at polymorphism A and allele B1 at 30 polymorphism B correlates with increased milk production of a farm animal.

Such correlations can be exploited in several ways. In the case of a strong correlation between a set of one or more polymorphic forms and a disease for which treatment is

available, detection of the polymorphic form set in a human or animal patient may justify immediate administration of treatment, or at least the institution of regular monitoring of the patient. Detection of a polymorphic form correlated with serious disease in a couple contemplating a family may also be valuable to the couple in their reproductive decisions.

5 For example, the female partner might elect to undergo in vitro fertilization to avoid the possibility of transmitting such a polymorphism from her husband to her offspring. In the case of a weaker, but still statistically significant correlation between a polymorphic set and human disease, immediate therapeutic intervention or monitoring may not be justified. Nevertheless, the patient can be motivated to begin simple life-style changes (e.g., diet, exercise) that can be accomplished at little cost to the patient but confer potential benefits in reducing the risk of conditions to which the patient may have increased susceptibility by virtue of variant alleles. Identification of a polymorphic set in a patient correlated with enhanced receptiveness to one of several treatment regimes for a disease indicates that this treatment regime should be followed.

15 For animals and plants, correlations between characteristics and phenotype are useful for breeding for desired characteristics. For example, Beitz et al., U.S. Pat. No. 5,292,639 discuss use of bovine mitochondrial polymorphisms in a breeding program to improve milk production in cows. To evaluate the effect of mtDNA D-loop sequence polymorphism on milk production, each cow was assigned a value of 1 if variant or 0 if wild type with respect to a prototypical mitochondrial DNA sequence at each of 17 locations considered.

The previous section concerns identifying correlations between phenotypic traits and polymorphisms that directly or indirectly contribute to those traits. The present section describes identification of a physical linkage between a genetic locus associated with a trait of interest and polymorphic markers that are not associated with the trait, but are in physical proximity with the genetic locus responsible for the trait and co-segregate with it. Such analysis is useful for mapping a genetic locus associated with a phenotypic trait to a chromosomal position, and thereby cloning gene(s) responsible for the trait. See Lander et al., *Proc. Natl. Acad. Sci. (USA)* 83, 7353-7357 (1986); Lander et al., *Proc. Natl. Acad. Sci. (USA)* 84, 2363-2367 (1987); Donis-Keller et al., *Cell* 51, 319-337 (1987); Lander et al., 25 *Genetics* 121, 185-199 (1989)). Genes localized by linkage can be cloned by a process known as directional cloning. See Wainwright, *Med. J. Australia* 159, 170-174 (1993); Collins, *Nature Genetics* 1, 3-6 (1992) (each of which is incorporated by reference in its entirety for all purposes).

Linkage studies are typically performed on members of a family. Available members of the family are characterized for the presence or absence of a phenotypic trait and for a set of polymorphic markers. The distribution of polymorphic markers in an informative meiosis is then analyzed to determine which polymorphic markers co-segregate with a phenotypic trait. See, e.g., Kerem et al., *Science* 245, 1073-1080 (1989); Monaco et al., *Nature* 316, 842 (1985); Yamoka et al., *Neurology* 40, 222-226 (1990); Rossiter et al., *FASEB Journal* 5, 21-27 (1991).

Linkage is analyzed by calculation of LOD (log of the odds) values. A lod value is the relative likelihood of obtaining observed segregation data for a marker and a genetic locus when the two are located at a recombination fraction  $\theta$ , versus the situation in which the two are not linked, and thus segregating independently (Thompson & Thompson, *Genetics in Medicine* (5th ed, W.B. Saunders Company, Philadelphia, 1991); Strachan, "Mapping the human genome" in *The Human Genome* (BIOS Scientific Publishers Ltd, Oxford), Chapter 4). A series of likelihood ratios are calculated at various recombination fractions ( $\theta$ ), ranging from  $\theta = 0.0$  (coincident loci) to  $\theta = 0.50$  (unlinked). Thus, the likelihood at a given value of  $\theta$  is: probability of data if loci linked at  $\theta$  to probability of data if loci unlinked. The computed likelihood is usually expressed as the  $\log_{10}$  of this ratio (i.e., a lod score). For example, a lod score of 3 indicates 1000:1 odds against an apparent observed linkage being a coincidence. The use of logarithms allows data collected from different families to be combined by simple addition. Computer programs are available for the calculation of lod scores for differing values of  $\theta$  (e.g., LIPED, MLINK (Lathrop, *Proc. Nat. Acad. Sci. (USA)* 81, 3443-3446 (1984)). For any particular lod score, a recombination fraction may be determined from mathematical tables. See Smith et al., *Mathematical tables for research workers in human genetics* (Churchill, London, 1961); Smith, *Ann. Hum. Genet.* 32, 127-150 (1968). The value of  $\theta$  at which the lod score is the highest is considered to be the best estimate of the recombination fraction.

Positive lod score values suggest that the two loci are linked, whereas negative values suggest that linkage is less likely (at that value of  $\theta$ ) than the possibility that the two loci are unlinked. By convention, a combined lod score of + 3 or greater (equivalent to greater than 1000:1 odds in favor of linkage) is considered definitive evidence that two loci are linked. Similarly, by convention, a negative lod score of -2 or less is taken as definitive evidence against linkage of the two loci being compared. Negative linkage data are useful in

excluding a chromosome or a segment thereof from consideration. The search focuses on the remaining non-excluded chromosomal locations.

The invention further provides transgenic nonhuman animals capable of expressing an exogenous variant gene and/or having one or both alleles of an endogenous variant gene  
5 inactivated. Expression of an exogenous variant gene is usually achieved by operably linking the gene to a promoter and optionally an enhancer, and microinjecting the construct into a zygote. See Hogan et al., "Manipulating the Mouse Embryo, A Laboratory Manual," Cold Spring Harbor Laboratory. (1989). Inactivation of endogenous variant genes can be achieved by forming a transgene in which a cloned variant gene is inactivated by insertion of  
10 a positive selection marker. See Capecchi, Science 244, 1288-1292. The transgene is then introduced into an embryonic stem cell, where it undergoes homologous recombination with an endogenous variant gene. Mice and other rodents are preferred animals. Such animals provide useful drug screening systems.

The invention further provides methods for assessing the pharmacogenomic  
15 susceptibility of a subject harboring a single nucleotide polymorphism to a particular pharmaceutical compound, or to a class of such compounds. Genetic polymorphism in drug-metabolizing enzymes, drug transporters, receptors for pharmaceutical agents, and other drug targets have been correlated with individual differences based on distinction in the efficacy and toxicity of the pharmaceutical agent administered to a subject. Pharmacogenomic  
20 characterization of a subjects susceptibility to a drug enhances the ability to tailor a dosing regimen to the particular genetic constitution of the subject, thereby enhancing and optimizing the therapeutic effectiveness of the therapy.

In cases in which a cSNP leads to a polymorphic protein that is ascribed to be the cause of a pathological condition, method of treating such a condition includes administering  
25 to a subject experiencing the pathology the wild type cognate of the polymorphic protein. Once administered in an effective dosing regimen, the wild type cognate provides complementation or remediation of the defect due to the polymorphic protein. The subject's condition is ameliorated by this protein therapy.

A subject suspected of suffering from a pathology ascribable to a polymorphic protein  
30 that arises from a cSNP is to be diagnosed using any of a variety of diagnostic methods capable of identifying the presence of the cSNP in the nucleic acid, or of the cognate

polymorphic protein, in a suitable clinical sample taken from the subject. Once the presence of the cSNP has been ascertained, and the pathology is correctable by administering a normal or wild-type gene, the subject is treated with a pharmaceutical composition that includes a nucleic acid that harbors the correcting wild-type gene, or a fragment containing a correcting sequence of the wild-type gene. Non-limiting examples of ways in which such a nucleic acid may be administered include incorporating the wild-type gene in a viral vector, such as an adenovirus or adeno associated virus, and administration of a naked DNA in a pharmaceutical composition that promotes intracellular uptake of the administered nucleic acid. Once the nucleic acid that includes the gene coding for the wild-type allele of the polymorphism is incorporated within a cell of the subject, it will initiate *de novo* biosynthesis of the wild-type gene product. If the nucleic acid is further incorporated into the genome of the subject, the treatment will have long-term effects, providing *de novo* synthesis of the wild-type protein for a prolonged duration. The synthesis of the wild-type protein in the cells of the subject will contribute to a therapeutic enhancement of the clinical condition of the subject.

A subject suffering from a pathology ascribed to a SNP may be treated so as to correct the genetic defect. (See Kren et al., Proc. Natl. Acad. Sci. USA 96:10349-10354 (1999)). Such a subject is identified by any method that can detect the polymorphism in a sample drawn from the subject. Such a genetic defect may be permanently corrected by administering to such a subject a nucleic acid fragment incorporating a repair sequence that supplies the wild-type nucleotide at the position of the SNP. This site-specific repair sequence encompasses an RNA/DNA oligonucleotide which operates to promote endogenous repair of a subject's genomic DNA. Upon administration in an appropriate vehicle, such as a complex with polyethylenimine or encapsulated in anionic liposomes, a genetic defect leading to an inborn pathology may be overcome, as the chimeric oligonucleotides induces incorporation of the wild-type sequence into the subject's genome. Upon incorporation, the wild-type gene product is expressed, and the replacement is propagated, thereby engendering a permanent repair.

The invention further provides kits comprising at least one allele-specific oligonucleotide as described above. Often, the kits contain one or more pairs of allele-specific oligonucleotides hybridizing to different forms of a polymorphism. In some kits, the allele-specific oligonucleotides are provided immobilized to a substrate. For example, the same substrate can comprise allele-specific oligonucleotide probes for detecting at least 10, 100, 1000 or all of the polymorphisms shown in the Table. Optional additional

components of the kit include, for example, restriction enzymes, reverse-transcriptase or polymerase, the substrate nucleoside triphosphates, means used to label (for example, an avidin-enzyme conjugate and enzyme substrate and chromogen if the label is biotin), and the appropriate buffers for reverse transcription, PCR, or hybridization reactions. Usually, the 5 kit also contains instructions for carrying out the hybridizing methods.

Several aspects of the present invention rely on having available the polymorphic proteins encoded by the nucleic acids comprising a SNP of the inventions. There are various methods of isolating these nucleic acid sequences. For example, DNA is isolated from a genomic or cDNA library using labeled oligonucleotide probes having sequences 10 complementary to the sequences disclosed herein.

Such probes can be used directly in hybridization assays. Alternatively probes can be designed for use in amplification techniques such as PCR.

To prepare a cDNA library, mRNA is isolated from tissue such as heart or pancreas, preferably a tissue wherein expression of the gene or gene family is likely to occur. cDNA is 15 prepared from the mRNA and ligated into a recombinant vector. The vector is transfected into a recombinant host for propagation, screening and cloning. Methods for making and screening cDNA libraries are well known, See Gubler, U. and Hoffman, B.J. Gene 25:263-269 (1983) and Sambrook et al.

For a genomic library, for example, the DNA is extracted from tissue and either 20 mechanically sheared or enzymatically digested to yield fragments of about 12-20 kb. The fragments are then separated by gradient centrifugation from undesired sizes and are constructed in bacteriophage lambda vectors. These vectors and phage are packaged *in vitro*, as described in Sambrook, et al. Recombinant phage are analyzed by plaque hybridization as described in Benton and Davis, Science 196:180-182 (1977). Colony hybridization is carried 25 out as generally described in M. Grunstein et al. Proc. Natl. Acad. Sci. USA. 72:3961-3965 (1975). DNA of interest is identified in either cDNA or genomic libraries by its ability to hybridize with nucleic acid probes, for example on Southern blots, and these DNA regions are isolated by standard methods familiar to those of skill in the art. See Sambrook, et al.

In PCR techniques, oligonucleotide primers complementary to the two 3' borders of 30 the DNA region to be amplified are synthesized. The polymerase chain reaction is then carried out using the two primers. See PCR Protocols: a Guide to Methods and Applications

(Innis, M., Gelfand, D., Sninsky, J. and White, T., eds.), Academic Press, San Diego (1990).

Primers can be selected to amplify the entire regions encoding a full-length sequence of interest or to amplify smaller DNA segments as desired. PCR can be used in a variety of protocols to isolate cDNA's encoding a sequence of interest. In these protocols, appropriate

5 primers and probes for amplifying DNA encoding a sequence of interest are generated from analysis of the DNA sequences listed herein. Once such regions are PCR-amplified, they can be sequenced and oligonucleotide probes can be prepared from the sequence.

Once DNA encoding a sequence comprising a cSNP is isolated and cloned, one can express the encoded polymorphic proteins in a variety of recombinantly engineered cells. It

10 is expected that those of skill in the art are knowledgeable in the numerous expression systems available for expression of DNA encoding a sequence of interest. No attempt to describe in detail the various methods known for the expression of proteins in prokaryotes or eukaryotes is made here.

In brief summary, the expression of natural or synthetic nucleic acids encoding a  
15 sequence of interest will typically be achieved by operably linking the DNA or cDNA to a promoter (which is either constitutive or inducible), followed by incorporation into an expression vector. The vectors can be suitable for replication and integration in either prokaryotes or eukaryotes. Typical expression vectors contain, initiation sequences, transcription and translation terminators, and promoters useful for regulation of the  
20 expression of a polynucleotide sequence of interest. To obtain high level expression of a cloned gene, it is desirable to construct expression plasmids which contain, at the minimum, a strong promoter to direct transcription, a ribosome binding site for translational initiation, and a transcription/translation terminator. The expression vectors may also comprise generic expression cassettes containing at least one independent terminator sequence, sequences  
25 permitting replication of the plasmid in both eukaryotes and prokaryotes, i.e., shuttle vectors, and selection markers for both prokaryotic and eukaryotic systems. See Sambrook et al.

A variety of prokaryotic expression systems may be used to express the polymorphic proteins of the invention. Examples include *E. coli*, *Bacillus*, *Streptomyces*, and the like.

It is preferred to construct expression plasmids which contain, at the minimum, a  
30 strong promoter to direct transcription, a ribosome binding site for translational initiation, and a transcription/translation terminator. Examples of regulatory regions suitable for this

purpose in *E. coli* are the promoter and operator region of the *E. coli* tryptophan biosynthetic pathway as described by Yanofsky, C., J. Bacterial. 158:1018-1024 (1984) and the leftward promoter of phage lambda (P<sub>l</sub>) as described by A, I. and Hagen, D., Ann. Rev. Genet. 14:399-445 (1980). The inclusion of selection markers in DNA vectors transformed in *E. coli* is also useful. Examples of such markers include genes specifying resistance to ampicillin, tetracycline, or chloramphenicol. See Sambrook et al. for details concerning selection markers for use in *E. coli*.

To enhance proper folding of the expressed recombinant protein, during purification from *E. coli*, the expressed protein may first be denatured and then renatured. This can be accomplished by solubilizing the bacterially produced proteins in a chaotropic agent such as guanidine HCl and reducing all the cysteine residues with a reducing agent such as beta-mercaptoethanol. The protein is then renatured, either by slow dialysis or by gel filtration. See U.S. Patent No. 4,511,503. Detection of the expressed antigen is achieved by methods known in the art as radioimmunoassay, or Western blotting techniques or immunoprecipitation. Purification from *E. coli* can be achieved following procedures such as those described in U.S. Patent No. 4,511,503.

Any of a variety of eukaryotic expression systems such as yeast, insect cell lines, bird, fish, and mammalian cells, may also be used to express a polymorphic protein of the invention. As explained briefly below, a nucleotide sequence harboring a cSNP may be expressed in these eukaryotic systems. Synthesis of heterologous proteins in yeast is well known. Methods in Yeast Genetics, Sherman, F., et al., Cold Spring Harbor Laboratory, (1982) is a well recognized work describing the various methods available to produce the protein in yeast. Suitable vectors usually have expression control sequences, such as promoters, including 3-phosphoglycerate kinase or other glycolytic enzymes, and an origin of replication, termination sequences and the like as desired. For instance, suitable vectors are described in the literature (Botstein, et al., Gene 8:17-24 (1979); Broach, et al., Gene 8:121-133 (1979)).

Two procedures are used in transforming yeast cells. In one case, yeast cells are first converted into protoplasts using zymolyase, lyticase or glusulase, followed by addition of DNA and polyethylene glycol (PEG). The PEG-treated protoplasts are then regenerated in a 3% agar medium under selective conditions. Details of this procedure are given in the papers by J.D. Beggs, Nature (London) 275:104-109 (1978); and Hinnen, A., et al., Proc. Natl.

Acad. Sci. USA, 75:1929-1933 (1978). The second procedure does not involve removal of the cell wall. Instead the cells are treated with lithium chloride or acetate and PEG and put on selective plates (Ito, H., et al., J. Bact, 153:163-168 (1983)). cells and applying standard protein isolation techniques to the lysates:

5        The purification process can be monitored by using Western blot techniques or radioimmunoassay or other standard techniques. The sequences encoding the proteins of the invention can also be ligated to various immunoassay expression vectors for use in transforming cell cultures of, for instance, mammalian, insect, bird or fish origin. Illustrative of cell cultures useful for the production of the polypeptides are mammalian cells.

10      Mammalian cell systems often will be in the form of monolayers of cells although mammalian cell suspensions may also be used. A number of suitable host cell lines capable of expressing intact proteins have been developed in the art, and include the HEK293, BHK21, and CHO cell lines, and various human cells such as COS cell lines, HeLa cells, myeloma cell lines, Jurkat cells, etc. Expression vectors for these cells can include

15      expression control sequences, such as an origin of replication, a promoter (e.g., the CMV promoter, a HSV *tk* promoter or *pgk* (phosphoglycerate kinase) promoter), an enhancer (Queen et al. Immunol. Rev., 89:49 (1986)) and necessary processing information sites, such as ribosome binding sites, RNA splice sites, polyadenylation sites (e.g., an SV40 large T Ag poly A addition site), and transcriptional terminator sequences.

20      Other animal cells are available, for instance, from the American Type Culture Collection Catalogue of Cell Lines and Hybridomas (7th edition, (1992)). Appropriate vectors for expressing the proteins of the invention in insect cells are usually derived from baculovirus. Insect cell lines include mosquito larvae, silkworm, armyworm, moth and Drosophila cell lines such as a Schneider cell line (See Schneider J. Embryol. Exp. Morphol., 27:353-365 (1987). As indicated above, the vector, e.g., a plasmid, which is used to transform the host cell, preferably contains DNA sequences to initiate transcription and sequences to control the translation of the protein. These sequences are referred to as expression control sequences. As with yeast, when higher animal host cells are employed, polyadenylation or transcription terminator sequences from known mammalian genes need to

25      be incorporated into the vector. An example of a terminator sequence is the polyadenylation sequence from the bovine growth hormone gene. Sequences for accurate splicing of the transcript may also be included. An example of a splicing sequence is the VP1 intron from SV40 (Sprague, J. et al., J. Virol. 45: 773-781 (1983)). Additionally, gene sequences to

30      48

control replication in the host cell may be Saveria-Campo, M., 1985, "Bovine Papilloma virus DNA a Eukaryotic Cloning Vector" in DNA Cloning Vol. II a Practical Approach Ed. D.M. Glover, IRL Press, Arlington, Virginia pp. 213-238. The host cells are competent or rendered competent for transformation by various means. There are several well-known 5 methods of introducing DNA into animal cells. These include: calcium phosphate precipitation, fusion of the recipient cells with bacterial protoplasts containing the DNA, treatment of the recipient cells with liposomes containing the DNA, DEAE dextran, electroporation and micro-injection of the DNA directly into the cells.

The transformed cells are cultured by means well known in the art (Biochemical 10 Methods in Cell Culture and Virology, Kuchler, R.J., Dowden, Hutchinson and Ross, Inc., (1977)). The expressed polypeptides are isolated from cells grown as suspensions or as monolayers. The latter are recovered by well known mechanical, chemical or enzymatic means.

General methods of expressing recombinant proteins are also known and are 15 exemplified in R. Kaufman, Methods in Enzymology 185, 537-566 (1990). As defined herein "operably linked" refers to linkage of a promoter upstream from a DNA sequence such that the promoter mediates transcription of the DNA sequence. Specifically, "operably linked" means that the isolated polynucleotide of the invention and an expression control sequence are situated within a vector or cell in such a way that the gene encoding the protein 20 is expressed by a host cell which has been transformed (transfected) with the ligated polynucleotide/expression sequence. The term "vector", refers to viral expression systems, autonomous self-replicating circular DNA (plasmids), and includes both expression and nonexpression plasmids.

The term "gene" as used herein is intended to refer to a nucleic acid sequence which 25 encodes a polypeptide. This definition includes various sequence polymorphisms, mutations, and/or sequence variants wherein such alterations do not affect the function of the gene product. The term "gene" is intended to include not only coding sequences but also regulatory regions such as promoters, enhancers, termination regions and similar untranslated nucleotide sequences. The term further includes all introns and other DNA sequences spliced 30 from the mRNA transcript, along with variants resulting from alternative splice sites.

A number of types of cells may act as suitable host cells for expression of the protein. Mammalian host cells include, for example, monkey COS cells, Chinese Hamster Ovary (CHO) cells, human kidney 293 cells, human epidermal A43 1 cells, human Co10205 cells, 3T3 cells, CV-1 cells, other transformed primate cell lines, normal diploid cells, cell strains derived from in vitro culture of primary tissue, primary explants, HeLa cells, mouse L cells, BHK, HL- 60, U937, HaK or Jurkat cells. Alternatively, it may be possible to produce the protein in lower eukaryotes such as yeast or in prokaryotes such as bacteria. Potentially suitable yeast strains include *Saccharomyces cerevisiae*, *Schizosaccharomyces pombe*, *Kluyveromyces* strains, *Candida* or any yeast strain capable of expressing heterologous proteins. Potentially suitable bacterial strains include *Escherichia coli*, *Bacillus subtilis*, *Salmonella typhimurium*, or any bacterial strain capable of expressing heterologous proteins. If the protein is made in yeast or bacteria, it may be necessary to modify the protein produced therein, for example by phosphorylation or glycosylation of the appropriate sites, in order to obtain the functional protein.

The protein may also be produced by operably linking the isolated polynucleotide of the invention to suitable control sequences in one or more insect expression vectors, and employing an insect expression system. Materials and methods for baculovirus/insect cell expression systems are commercially available in kit form from, e.g., Invitrogen, San Diego, California, U.S.A. (the MaxBac<sup>©</sup> kit), and such methods are well known in the art, as described in Summers and Smith, Texas Agricultural Experiment Station Bulletin No. 1555 (1987), incorporated herein by reference. As used herein, an insect cell capable of expressing a polynucleotide of the present invention is "transformed." The protein of the invention may be prepared by culturing transformed host cells under culture conditions suitable to express the recombinant protein.

The polymorphic protein of the invention may also be expressed as a product of transgenic animals, e.g., as a component of the milk of transgenic cows, goats, pigs, or sheep which are characterized by somatic or germ cells containing a nucleotide sequence encoding the protein. The protein may also be produced by known conventional chemical synthesis. Methods for constructing the proteins of the present invention by synthetic means are known to those skilled in the art.

The polymorphic proteins produced by recombinant DNA technology may be purified by techniques commonly employed to isolate or purify recombinant proteins. Recombinantly

produced proteins can be directly expressed or expressed as a fusion protein. The protein is then purified by a combination of cell lysis (e.g., sonication) and affinity chromatography. For fusion products, subsequent digestion of the fusion protein with an appropriate proteolytic enzyme releases the desired polypeptide. The polypeptides of this invention may 5 be purified to substantial purity by standard techniques well known in the art, including selective precipitation with such substances as ammonium sulfate, column chromatography, immunopurification methods, and others. See, for instance, R. Scopes, *Protein Purification: Principles and Practice*, Springer-Verlag: New York (1982), incorporated herein by reference. For example, in an embodiment, antibodies may be raised to the proteins of the invention as 10 described herein. Cell membranes are isolated from a cell line expressing the recombinant protein, the protein is extracted from the membranes and immunoprecipitated. The proteins may then be further purified by standard protein chemistry techniques as described above.

The resulting expressed protein may then be purified from such culture (i.e., from culture medium or cell extracts) using known purification processes, such as gel filtration 15 and ion exchange chromatography. The purification of the protein may also include an affinity column containing agents which will bind to the protein; one or more column steps over such affinity resins as concanavalin A-agarose, heparin-Toyopearl@ or Cibacrom blue 3GA Sepharose B; one or more steps involving hydrophobic interaction chromatography using such resins as phenyl ether, butyl ether, or propyl ether; or immunoaffinity 20 chromatography. Alternatively, the protein of the invention may also be expressed in a form which will facilitate purification. For example, it may be expressed as a fusion protein, such as those of maltose binding protein (MBP), glutathione-S-transferase (GST) or thioredoxin (TRX). Kits for expression and purification of such fusion proteins are commercially available from New England BioLab (Beverly, MA), Pharmacia (Piscataway, NJ) and 25 InVitrogen, respectively. The protein can also be tagged with an epitope and subsequently purified by using a specific antibody directed to such epitope. One such epitope ("Flag") is commercially available from Kodak (New Haven, CT). Finally, one or more reverse-phase high performance liquid chromatography (RP- HPLC) steps employing hydrophobic RP- HPLC media, e.g., silica gel having pendant methyl or other aliphatic groups, can be 30 employed to further purify the protein. Some or all of the foregoing purification steps, in various combinations, can also be employed to provide a substantially homogeneous isolated recombinant protein. The protein thus purified is substantially free of other

mammalian proteins and is defined in accordance with the present invention as an "isolated protein."

The term "antibody" as used herein refers to immunoglobulin molecules and immunologically active portions of immunoglobulin molecules, *i.e.*, molecules that contain an antigen binding site that specifically binds (immunoreacts with) an antigen, such as polymorphic. Such antibodies include, but are not limited to, polyclonal, monoclonal, chimeric, single chain, F<sub>ab</sub> and F<sub>(ab)2</sub> fragments, and an F<sub>ab</sub> expression library. In a specific embodiment, antibodies to human polymorphic proteins are disclosed.

The phrase "specifically binds to", "immunospecifically binds to" or is "specifically immunoreactive with", an antibody when referring to a protein or peptide, refers to a binding reaction which is determinative of the presence of the protein in the presence of a heterogeneous population of proteins and other biological materials. Thus, for example, under designated immunoassay conditions, the specified antibodies bind to a particular protein and do not bind in a significant amount to other proteins present in the sample.

Specific binding to an antibody under such conditions may require an antibody that is selected for its specificity for a particular protein. Of particular interest in the present invention is an antibody that binds immunospecifically to a polymorphic protein but not to its cognate wild type allelic protein, or vice versa. A variety of immunoassay formats may be used to select antibodies specifically immunoreactive with a particular protein. For example, solid-phase ELISA immunoassays are routinely used to select monoclonal antibodies specifically immunoreactive with a protein. See Harlow and Lane (1988) *Antibodies, a Laboratory Manual*, Cold Spring Harbor Publications, New York, for a description of immunoassay formats and conditions that can be used to determine specific immunoreactivity.

Polyclonal and/or monoclonal antibodies that immunospecifically bind to polymorphic gene products but not to the corresponding prototypical or "wild-type" gene products are also provided. Antibodies can be made by injecting mice or other animals with the variant gene product or synthetic peptide. Monoclonal antibodies are screened as are described, for example, in Harlow & Lane, *Antibodies, A Laboratory Manual*, Cold Spring Harbor Press, New York (1988); Goding, *Monoclonal antibodies, Principles and Practice* (2d ed.) Academic Press, New York (1986). Monoclonal antibodies are tested for specific

immunoreactivity with a variant gene product and lack of immunoreactivity to the corresponding prototypical gene product.

An isolated polymorphic protein, or a portion or fragment thereof, can be used as an immunogen to generate the antibody that bind the polymorphic protein using standard techniques for polyclonal and monoclonal antibody preparation. The full-length polymorphic protein can be used or, alternatively, the invention provides antigenic peptide fragments of polymorphic for use as immunogens. The antigenic peptide of a polymorphic protein of the invention comprises at least 8 amino acid residues of the amino acid sequence encompassing the polymorphic amino acid and encompasses an epitope of the polymorphic protein such that an antibody raised against the peptide forms a specific immune complex with the polymorphic protein. Preferably, the antigenic peptide comprises at least 10 amino acid residues, more preferably at least 15 amino acid residues, even more preferably at least 20 amino acid residues, and most preferably at least 30 amino acid residues. Preferred epitopes encompassed by the antigenic peptide are regions of polymorphic that are located on the surface of the protein, e.g., hydrophilic regions.

For the production of polyclonal antibodies, various suitable host animals (e.g., rabbit, goat, mouse or other mammal) may be immunized by injection with the polymorphic protein. An appropriate immunogenic preparation can contain, for example, recombinantly expressed polymorphic protein or a chemically synthesized polymorphic polypeptide. The preparation can further include an adjuvant. Various adjuvants used to increase the immunological response include, but are not limited to, Freund's (complete and incomplete), mineral gels (e.g., aluminum hydroxide), surface active substances (e.g., lysolecithin, pluronic polyols, polyanions, peptides, oil emulsions, dinitrophenol, etc.), human adjuvants such as *Bacille Calmette-Guerin* and *Corynebacterium parvum*, or similar immunostimulatory agents. If desired, the antibody molecules directed against polymorphic proteins can be isolated from the mammal (e.g., from the blood) and further purified by well known techniques, such as protein A chromatography, to obtain the IgG fraction.

The term "monoclonal antibody" or "monoclonal antibody composition", as used herein, refers to a population of antibody molecules that originates from the clone of a singly hybridoma cell, and that contains only one type of antigen binding site capable of immunoreacting with a particular epitope of a polymorphic protein. A monoclonal antibody composition thus typically displays a single binding affinity for a particular polymorphic

protein with which it immunoreacts. For preparation of monoclonal antibodies directed towards a particular polymorphic protein, or derivatives, fragments, analogs or homologs thereof, any technique that provides for the production of antibody molecules by continuous cell line culture may be utilized. Such techniques include, but are not limited to, the hybridoma technique (see Kohler & Milstein, 1975 *Nature* 256: 495-497); the trioma technique; the human B-cell hybridoma technique (see Kozbor, *et al.*, 1983 *Immunol Today* 4: 72) and the EBV hybridoma technique to produce human monoclonal antibodies (see Cole, *et al.*, 1985 In: MONOCLONAL ANTIBODIES AND CANCER THERAPY, Alan R. Liss, Inc., pp. 77-96). Human monoclonal antibodies may be utilized in the practice of the present invention and may be produced by using human hybridomas (see Cote, *et al.*, 1983. *Proc Natl Acad Sci USA* 80: 2026-2030) or by transforming human B-cells with Epstein Barr Virus *in vitro* (see Cole, *et al.*, 1985 In: MONOCLONAL ANTIBODIES AND CANCER THERAPY, Alan R. Liss, Inc., pp. 77-96).

According to the invention, techniques can be adapted for the production of single-chain antibodies specific to a polymorphic protein (see e.g., U.S. Patent No. 4,946,778). In addition, methodologies can be adapted for the construction of F<sub>ab</sub> expression libraries (see e.g., Huse, *et al.*, 1989 *Science* 246: 1275-1281) to allow rapid and effective identification of monoclonal F<sub>ab</sub> fragments with the desired specificity for a polymorphic protein or derivatives, fragments, analogs or homologs thereof. Non-human antibodies can be "humanized" by techniques well known in the art. See e.g., U.S. Patent No. 5,225,539. Antibody fragments that contain the idiotypes to a polymorphic protein may be produced by techniques known in the art including, but not limited to: (i) an F<sub>(ab')2</sub> fragment produced by pepsin digestion of an antibody molecule; (ii) an F<sub>ab</sub> fragment generated by reducing the disulfide bridges of an F<sub>(ab')2</sub> fragment; (iii) an F<sub>ab</sub> fragment generated by the treatment of the antibody molecule with papain and a reducing agent and (iv) F<sub>v</sub> fragments.

Additionally, recombinant anti-polymorphic protein antibodies, such as chimeric and humanized monoclonal antibodies, comprising both human and non-human portions, which can be made using standard recombinant DNA techniques, are within the scope of the invention. Such chimeric and humanized monoclonal antibodies can be produced by recombinant DNA techniques known in the art, for example using methods described in PCT International Application No. PCT/US86/02269; European Patent Application No. 184,187; European Patent Application No. 171,496; European Patent Application No. 173,494; PCT International Publication No. WO 86/01533; U.S. Pat. No. 4,816,567; European Patent

Application No. 125,023; Better *et al.* (1988) *Science* 240:1041-1043; Liu *et al.* (1987) *PNAS* 84:3439-3443; Liu *et al.* (1987) *J Immunol.* 139:3521-3526; Sun *et al.* (1987) *PNAS* 84:214-218; Nishimura *et al.* (1987) *Cancer Res* 47:999-1005; Wood *et al.* (1985) *Nature* 314:446-449; Shaw *et al.* (1988) *J Natl Cancer Inst* 80:1553-1559; Morrison (1985) *Science* 229:1202-1207; Oi *et al.* (1986) *BioTechniques* 4:214; U.S. Pat. No. 5,225,539; Jones *et al.* (1986) *Nature* 321:552-525; Verhoeven *et al.* (1988) *Science* 239:1534; and Beidler *et al.* (1988) *J Immunol* 141:4053-4060.

In one embodiment, methodologies for the screening of antibodies that possess the desired specificity include, but are not limited to, enzyme-linked immunosorbent assay (ELISA) and other immunologically-mediated techniques known within the art.

Anti-polymorphic protein antibodies may be used in methods known within the art relating to the detection, quantitation and/or cellular or tissue localization of a polymorphic protein (*e.g.*, for use in measuring levels of the polymorphic protein within appropriate physiological samples, for use in diagnostic methods, for use in imaging the protein, and the like). In a given embodiment, antibodies for polymorphic proteins, or derivatives, fragments, analogs or homologs thereof, that contain the antibody-derived CDR, are utilized as pharmacologically-active compounds in therapeutic applications intended to treat a pathology in a subject that arises from the presence of the cSNP allele in the subject.

An anti-polymorphic protein antibody (*e.g.*, monoclonal antibody) can be used to isolate polymorphic proteins by a variety of immunochemical techniques, such as immunoaffinity chromatography or immunoprecipitation. An anti-polymorphic protein antibody can facilitate the purification of natural polymorphic protein from cells and of recombinantly produced polymorphic proteins expressed in host cells. Moreover, an anti-polymorphic protein antibody can be used to detect polymorphic protein (*e.g.*, in a cellular lysate or cell supernatant) in order to evaluate the abundance and pattern of expression of the polymorphic protein. Anti-polymorphic antibodies can be used diagnostically to monitor protein levels in tissue as part of a clinical testing procedure, *e.g.*, to, for example, determine the efficacy of a given treatment regimen. Detection can be facilitated by coupling (*i.e.*, physically linking) the antibody to a detectable substance. Examples of detectable substances include various enzymes, prosthetic groups, fluorescent materials, luminescent materials, bioluminescent materials, and radioactive materials. Examples of suitable enzymes include horseradish peroxidase, alkaline phosphatase,

-g αlactosidase, or acetylcholinesterase; examples of suitable prosthetic group complexes include streptavidin/biotin and avidin/biotin; examples of suitable fluorescent materials include umbelliferone, fluorescein, fluorescein isothiocyanate, rhodamine, dichlorotriazinylamine fluorescein, dansyl chloride or phycoerythrin; an example of a 5 luminescent material includes luminol; examples of bioluminescent materials include luciferase, luciferin, and aequorin, and examples of suitable radioactive material include  $^{125}\text{I}$ ,  $^{131}\text{I}$ ,  $^{35}\text{S}$  or  $^3\text{H}$ .

| Seq ID | CuraGen sequence ID | Base pos. of SNP | Polymorphic sequence   | Base before | Base after | Amino acid before | Amino acid after | Type of change | Protein classification of CuraGen gene | Name of protein identified following a BLASTX analysis of the CuraGen sequence   | Similarity (pValue) following a BLASTX analysis | Map location |
|--------|---------------------|------------------|--|-------------|------------|-------------------|------------------|----------------|--|--|---|--------------|
| 1      | cg43333349          | 1008             | CGCTGACAGGGGA<br>GTCTGAGCCACA[A<br>G]ACCCGGCTCAC<br>CGAGTGACGCAC<br>G  | A           | G          | Gln               | Gln              | SILENT-CODING  | ATPase_associated                      | Human Gene SWISSPROT-ID:P20648 POTASSIUM-TRANSPORTING ATPASE ALPHA CHAIN (EC 3.6.1.36) (PROTON PUMP) (GASTRIC H+/K+ ATPASE ALPHA SUBUNIT) - HOMO SAPIENS (HUMAN), 1035 aa. | 0   | 19           |
| 2      | cg43931765          | 2296             | ATGGATAAGTCCTCAT<br>CTGGTTGGATGC[A<br>T]GTGTACTCGTTG<br>GCCTCGTTCAAGGT | A           | T          | Thr               | Thr              | SILENT-CODING  | cadherin                               | Human Gene SWISSPROT-ID:P18084 INTEGRIN BETA-5 SUBUNIT PRECURSOR - HOMO SAPIENS (HUMAN), 799 aa.   | 0   | 3            |

|   |            |      |  |   |   |     |               |          |  |   |    |
|---|------------|------|--|---|---|-----|---------------|----------|--|---|----|
| 3 | cg44130533 | 1832 | AATACAAAGCTGA<br>GTGGAGAGCAGTT<br>[G]GGTGAAAGAAGT<br>AIGGCATTCAGAAG<br>T | T | G | Val | SILENT-CODING | cadherin | Human Gene SWISSNEW-ID:P13591 NEURAL CELL ADHESION MOLECULE, 140 KD ISOFORM PRECURSOR (NCAM-140) (CD56 ANTIGEN) - HOMO SAPIENS (HUMAN), 848 aa. pcis:SWISSPROT-ID:P13591 NEURAL CELL ADHESION MOLECULE, 140 KD ISOFORM PRECURSOR (NCAM-140) (CD56 ANTIGEN) - HOMO SAPIENS (HUMAN), 848 aa. | 0 | 11 |
| 4 | cg34888922 | 2330 | TATTGTTATTATGT<br>ATTCTGTTAC[A/G<br>CTGTTCTGTGTCA<br>CTGCTAACAGAGAA      | A | G | Thr | SILENT-CODING | cadherin | Human Gene SWISSNEW-ID:Q08554 DESMOCOLLIN 1A/1B PRECURSOR (DESMOSOMAL GLYCOPROTEIN 2/3) (DG2/DG3) - HOMO SAPIENS (HUMAN), 894 aa. pcis:SWISSPROT-ID:Q08554 DESMOCOLLIN 1A/1B PRECURSOR (DESMOSOMAL GLYCOPROTEIN 2/3) (DG2 / DG3) - HOMO SAPIENS (HUMAN), 894 aa.                           | 0 | 18 |

|   |            |      |  |   |   |     |     |               |          |   |   |                      |
|---|------------|------|--|---|---|-----|-----|---------------|----------|---|---|----------------------|
| 5 | cg34888922 | 815  | CAAGGAGCATTGA<br>CCGTGAGAAATA[C<br>T]GAACAGTTGC<br>GTATATGGCTATG       | C | T | Tyr | Tyr | SILENT-CODING | cadherin | Human Gene SWISSPROT-ID:Q08554 DESMOCOLLIN 1A/1B PRECURSOR (DESMOSOMAL GLYCOPROTEIN 2/3) (DG2/DG3) - HOMO SAPIENS (HUMAN), 894 aa. pcis:SWISSPROT-ID:Q08554 DESMOCOLLIN 1A/1B PRECURSOR (DESMOSOMAL GLYCOPROTEIN 2/3) (DG2 / DG3) - HOMO SAPIENS (HUMAN), 894 aa. | 0 | 18                   |
| 6 | cg40310734 | 1172 | TGGCGTUGGTATT[G<br>GGGCATTCACTGT[G<br>CJGCTGTCACTGAC<br>GTCAACGGGA[TG  | G | C | Val | Val | SILENT-CODING | cadherin | Human Gene SWISSPROT-ID:P08514 PLATELET MEMBRANE GLYCOPROTEIN IIb PRECURSOR (GP1IB) (INTEGRIN ALPHA-IIb) (CD41) - HOMO SAPIENS (HUMAN), 1039 aa.  | 0 | 17<br>(17q21.3<br>2) |
| 7 | cg40310734 | 2243 | AGGGGGCTATGA<br>AGCACAGACTGGC[C<br>/G]GTGCACCTGCC<br>CCAGGGGGCCAC<br>T | C | G | Ala | Ala | SILENT-CODING | cadherin | Human Gene SWISSPROT-ID:P08514 PLATELET MEMBRANE GLYCOPROTEIN IIb PRECURSOR (GP1IB) (INTEGRIN ALPHA-IIb) (CD41) - HOMO SAPIENS (HUMAN), 1039 aa.  | 0 | 17<br>(17q21.3<br>2) |
| 8 | cg40310734 | 812  | GTACTTGAA[G<br>GGGCTTCAGCTC/C<br>G]GTGGTCACTCAG<br>GCCGGAGAGCTGG       | C | G | Ser | Ser | SILENT-CODING | cadherin | Human Gene SWISSPROT-ID:P08514 PLATELET MEMBRANE GLYCOPROTEIN IIb PRECURSOR (GP1IB) (INTEGRIN ALPHA-IIb) (CD41) - HOMO SAPIENS (HUMAN), 1039 aa.  | 0 | 17<br>(17q21.3<br>2) |

|    |            |      |  |   |   |     |     |               |           |  |           |               |
|----|------------|------|--|---|---|-----|-----|---------------|-----------|--|-----------|---------------|
| 9  | cg43331935 | 1922 | GTGACAAGTACTT<br>CATAGAGGATGGI<br>G/TTCGCCCTGGICAT<br>CCACAGCCTGGAC<br>T | G | T | Gly | Gly | SILENT-CODING | cadherin  | Human Gene SWISSPROT-ID:P2004 NEURAL CELL ADHESION MOLECULE LI PRECURSOR (N-CAM(L1)) - HOMO SAPIENS (HUMAN), 1257 aa.  | 0         | X             |
| 10 | cg42388009 | 383  | AAGGAAAAACAA<br>TGAAGAACCGAAL<br>CTTGAAGACGAAG<br>ACTCTGAGGGCTGA<br>GA   | C | T | Asn | Asn | SILENT-CODING | cadherin  | Human Gene SWISSPROT-ID:P21815 BONE SIALOPROTEIN II PRECURSOR (BSP II) (CELL-BINDING SIALOPROTEIN) (INTEGRIN-BINDING SIALOPROTEIN) - HOMO SAPIENS (HUMAN), 317 aa. | 7.00E-172 | 4             |
| 11 | cg42388009 | 389  | AAAACAATGAAGA<br>ACCGAACGAAGA<br>CTTGAAGACTCTG<br>AGGCTGAGAATA<br>CA     | C | T | Asp | Asp | SILENT-CODING | cadherin  | Human Gene SWISSPROT-ID:P21815 BONE SIALOPROTEIN II PRECURSOR (BSP II) (CELL-BINDING SIALOPROTEIN) (INTEGRIN-BINDING SIALOPROTEIN) - HOMO SAPIENS (HUMAN), 317 aa. | 7.00E-172 | 4             |
| 12 | cg44126574 | 1289 | AGAACGGCCAGCC<br>CCTGGGATCCTC<br>GJGGGGATGTCTTC<br>CTCAGGTCTACT          | C | G | Leu | Leu | SILENT-CODING | cathepsin | Human Gene SPTRMBL-ID:Q64411 PROGASTRINSIN PRECURSOR (EC 3.4.23.3) (PEPSIN C) - CAVIA PORCELLUS (GUINEA PIG), 394 aa.  | 8.00E-155 | 6<br>(6p21.3) |

|    |            |      |   |   |   |     |     |               |            |  |           |               |
|----|------------|------|---|---|---|-----|-----|---------------|------------|--|-----------|---------------|
| 13 | cg43970983 | 3066 | GGACTCCAGTGTCC<br>CAGGGCATCCAGIC<br>/TACATCTTATCC<br>TGGCGGCCACTCA        | C | T | Ser | Ser | SILENT-CODING | collagen   | Human Gene SWISSPROT-ID:Q02388 COLLAGEN ALPHA 1(VI) CHAIN PRECURSOR (LONG-CHAIN COLLAGEN) (LC COLLAGEN) - HOMO SAPIENS (HUMAN), 2944 aa. | 0         | 3<br>(3p21.3) |
| 14 | cg44032748 | 245  | TAAGACGGGCAGCA<br>TACACCCGGCAGCIA<br>/GIGTTACCTGCCA<br>GCTGAGCAACTGG<br>T | A | G | Ala | Ala | SILENT-CODING | complement | Human Gene SWISSPROT-ID:P07357 COMPLEMENT COMPONENT C8 ALPHA CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 584 aa.                             | 0         | 1<br>(1p32)   |
| 15 | cg41553795 | 222  | TCCAGCCCAAGCT<br>CAATTITGATGCT<br>/GJAGCAGTTGCA<br>GGGACCTGGCTCC          | T | G | Ala | Ala | SILENT-CODING | complement | Human Gene Homologous to SWISSPROT-ID:P07360 COMPLEMENT C8 GAMMA CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 202 aa.                         | 1.40E-104 | 9<br>(9q34.3) |
| 16 | cg43942011 | 1371 | AGCTAGGAGGGCT<br>TGGTCTCCAACIA<br>/GJCCTATTTTICA<br>TTCTCCACAGTGC         | A | G | Gly | Gly | SILENT-CODING | complement | Human Gene Similar to TREMBL NEW-ID:E246058 COMPLEMENT RECEPTOR 2-MUS MUSCULUS (MOUSE), 651 aa (fragment).                               | 1.10E-69  | 1<br>(1q32)   |
| 17 | cg21644442 | 1219 | AACAGCCGGCAGA<br>TGTAACIGGTACIA<br>/CIGCCTTGCCCCAG<br>GGTGGGGCCCCGTGA     | A | C | Thr | Thr | SILENT-CODING | csf        | Human Gene SWISSPROT-ID:P09603 MACROPHAGE COLONY STIMULATING FACTOR-1 PRECURSOR (CSF-1) (MCSF) - HOMO SAPIENS (HUMAN), 554 aa.           | 5.00E-304 | 1<br>(1p21)   |

|    |            |      |  |   |   |     |               |               |  |           |                 |
|----|------------|------|--|---|---|-----|---------------|---------------|--|-----------|-----------------|
| 18 | cg41533258 | 597  | TCCAGGGCGGGC<br>AGGAGGGTCCCTG<br>(A/GTTGCCCTCCAT<br>CTGCAGAGCTTCC    | G | A | Leu | SILENT-CODING | csf           | Human Gene Homologous to SWISSPROT-ID:P09919<br>GRANULOCYTE COLONY-STIMULATING FACTOR PRECURSOR (G-CSF)<br>(PLURIPOTENT) HOMO SAPIENS (HUMAN), 207 aa.   | 1.50E-107 | 17<br>(17q11.2) |
| 19 | cg43996714 | 1743 | ATGTGCCCACTGC<br>ATTGGGTTGCCA/<br>GJGGAGTTGATACT<br>GGTGGGATCACAG    | A | G | Pro | SILENT-CODING | dehydrogenase | Human Gene TREMBL NEW-ID:G2979625 PYRUVATE DEHYDROGENASE COMPLEX PROTEIN X SUBUNIT PRECURSOR - HOMO SAPIENS (HUMAN), 501 aa.   | 1.60E-266 | 11              |
| 20 | cg43259523 | 366  | CAGAAATATGGAGG<br>CACAGGGAGCTTCIA<br>/TTTTTTATCCACT<br>GTGCTCGTGATAG | A | T | Ser | SILENT-CODING | dehydrogenase | Human Gene SWISSPROT-ID:P45954 ACYL-COA DEHYDROGENASE, SHORT-BRANCHED CHAIN SPECIFIC PRECURSOR (EC 1.3.99.-)(SBCAD) (2-METHYL BRANCHED CHAIN ACYL-COA DEHYDROGENASE) (2-MBCAD) - HOMO SAPIENS (HUMAN), 432 aa. | 2.00E-229 | 10<br>(10q25)   |

|    |            |      |  |   |   |     |               |               |  |           |          |
|----|------------|------|--|---|---|-----|---------------|---------------|--|-----------|----------|
| 21 | cg43057018 | 1528 | GAATAAGAAATTCAATCTGGATGCACTTGGTGACCCATA<br>CCCTGCCTTTGAA               | C | T | Leu | SILENT-CODING | dehydrogenase | Human Gene SWISS-NEW-ID: P08319 ALCOHOL DEHYDROGENASE CLASS II PI CHAIN (EC 1.1.1.- HOMO SAPIENS (HUMAN), 391 aa.)<br>[pcls:SWISSPROT-ID:P08319 ALCOHOL DEHYDROGENASE CLASS II PI CHAIN (EC 1.1.1.- HOMO SAPIENS (HUMAN), 391 aa.) | 1.30E-209 | 4 (4q22) |
| 22 | cg1395871  | 430  | GTGCCCTCAGAGGG<br>GCCAGCAGGCAC<br>A/GGGAAAACCG<br>AAACCACCAAGGA<br>CT  | A | G | Thr | SILENT-CODING | dynen         | Human Gene Homologous to SPTREMBL-ID: Q92816 CYTOPLASMIC DYNENIN 3 HEAVY CHAIN - HOMO SAPIENS (HUMAN), 197 aa (fragment).  | 2.50E-103 |          |
| 23 | cg1395871  | 436  | CAGAGGGCCAGC<br>AGGCACAGGAAAI<br>A/G1ACCGAAACCA<br>CCAAGGACTTGGC<br>TA | A | G | Lys | SILENT-CODING | dynen         | Human Gene Homologous to SPTREMBL-ID: Q92816 CYTOPLASMIC DYNENIN 3 HEAVY CHAIN - HOMO SAPIENS (HUMAN), 197 aa (fragment).  | 2.50E-103 |          |
| 24 | cg1395871  | 542  | AGCAAATGGGAAAG<br>TTTTTAAAGGAAC<br>TTGGCTTCCTCTG<br>GTGCCTGGCTTG       | C | T | Leu | SILENT-CODING | dynen         | Human Gene Homologous to SPTREMBL-ID: Q92816 CYTOPLASMIC DYNENIN 3 HEAVY CHAIN - HOMO SAPIENS (HUMAN), 197 aa (fragment).  | 2.50E-103 |          |

|    |            |      |  |   |   |     |               |         |   |           |    |
|----|------------|------|--|---|---|-----|---------------|---------|---|-----------|----|
| 25 | cgi395871  | 571  | CTTCTTCTGGTGTCT<br>TGGGCTTGTCTTCCT<br>[GATGAATTCAACC<br>GGATTGAGTTGG   | C | T | Phe | SILENT-CODING | dynamin | Human Gene Homologous to<br>SPTREMBL-ID:Q92816<br>CYTOPLASMIC DYNENIN 3<br>HEAVY CHAIN - HOMO<br>SAPIENS (HUMAN), 197 aa<br>(fragment).   | 2.50E-103 |    |
| 26 | cgi3950268 | 1269 | AGCGGCCACCAT<br>GGCCCTAGGGTC[G<br> ATCAACAAAGTCC<br>AGCAGCAATCATG<br>G | G | A | Asp | SILENT-CODING | eph     | Human Gene TREMBLNEW-ID:G285466 HEAT SHOCK PROTEIN 75 - HOMO SAPIENS (HUMAN), 649 aa.   | 0         | 16 |
| 27 | cgi3918531 | 461  | CCGATGGCTATGA<br>GCAGGGCTGCTCG[C<br> T]GTTGCTATTGAA<br>CACCTGGACAAGA   | C | T | Arg | SILENT-CODING | eph     | Human Gene Homologous to<br>SWISSNEW-ID:Q52500<br>THERMOSOME SUBUNIT (HEAT-SHOCK PROTEIN)-PYROCOCCUS KODAKARAENSIS, 546 aa. pels:SWISSPROF-ID:Q52500<br>THERMOSOME SUBUNIT (HEAT-SHOCK PROTEIN)-PYROCOCCUS SP. (STRAIN KODI), 546 aa. | 1.00E-104 | 5  |

|    |            |      |  |   |   |     |     |                   |          |   |           |
|----|------------|------|--|---|---|-----|-----|-------------------|----------|---|-----------|
| 28 | cg43957743 | 1146 | CAAGGITCCCTAACT<br>AAAGTGGCAGTTC<br>TTTCAGGTCTACT<br>GGCTCCACCTCTC | C | T | Glu | Glu | SILENT-<br>CODING | esterase | Human Gene SWISSNEW -<br>ID:Q15166 SERUM<br>PARAOXONASE/ARYLESTERA-<br>SE 3 (EC 3.1.1.2) (EC 3.1.8.1)<br>(PON 3) (SERUM<br>ARYLDIAKYLPHOSPHATASE<br>3) (A-ESTERASE 3)<br>(AROMATIC ESTERASE 3) -<br>HOMO SAPIENS (HUMAN), 341<br>aa (fragment).lpcls:SWISSPROT-<br>II:Q15166 SERUM<br>PARAOXONASE/ARYLESTERA-<br>SE 3 (EC 3.1.1.2) (EC 3.1.8.1)<br>(PON 3) (SERUM<br>ARYLDIAKYLPHOSPHATASE<br>3) (A-ESTERASE 3)<br>(AROMATIC ESTERASE 3) -<br>HOMO SAPIENS (HUMAN), 341<br>aa (fragment). | 1.90E-178 |
|----|------------|------|--|---|---|-----|-----|-------------------|----------|---|-----------|

|    |            |      |  |   |   |     |     |               |          |  |           |          |
|----|------------|------|--|---|---|-----|-----|---------------|----------|--|-----------|----------|
| 29 | cg43319420 | 963  | TCACCCCTCAGGAG<br>GTGGCTGTTCTGC/<br>TGTCCACGACAAC<br>TACAGAAACAACC | C | T | Cys | Cys | SILENT-CODING | esterase | Human Gene Similar to<br>SWISSNEW-ID:Q23917 3',5'-<br>CYCLIC-NUCLEOTIDE<br>PHOSPHODIESTERASE REGA<br>(EC 3.1.4.17) (PDEASE REGA)-<br>DICTYOSTELIUM<br>DISCODEUM (SLIME MOLD),<br>793 aa.[pels:SWISSPROT-<br>ID:Q23917 3',5'-CYCLIC-<br>NUCLEOTIDE<br>PHOSPHODIESTERASE REGA<br>(EC 3.1.4.17) (PDEASE REGA)-<br>DICTYOSTELIUM<br>DISCODEUM (SLIME MOLD),<br>793 aa. | 3.30E-60  | 21       |
| 30 | cg3001932  | 1631 | TCTTCAACATCGTC<br>TATTGGCTTA[CT<br>TTATGTGAACAAA<br>ACATGGCTCCC    | C | T | Tyr | Tyr | SILENT-CODING | gaba     | Human Gene SWISSPROT-<br>ID:P47870 GAMMA-<br>AMINOBUTYRIC-ACID<br>RECEPTOR BETA2 SUBUNIT<br>PRECURSOR (GABA(A)<br>RECEPTOR) - HOMO SAPIENS<br>(HUMAN), 474 aa.   | 1.90E-256 | 5 (5q34) |
| 31 | cg43975899 | 370  | GGATTGGACAG<br>ACTCCTAGATGGIC<br>/T]TATGACAATCGC<br>CTGAGACCAAGGAT | C | T | Gly | Gly | SILENT-CODING | gaba     | Human Gene SWISSPROT-<br>ID:P14867 GAMMA-<br>AMINOBUTYRIC-ACID<br>RECEPTOR ALPHA1 SUBUNIT<br>PRECURSOR (GABA(A)<br>RECEPTOR) - HOMO SAPIENS<br>(HUMAN), 456 aa.  | 1.30E-248 | 5 (5q34) |

|    |            |      |   |   |   |     |     |               |               |  |           |                     |
|----|------------|------|---|---|---|-----|-----|---------------|---------------|--|-----------|---------------------|
| 32 | cg43299024 | 1643 | GGGCCCACTTCCC<br>CCTGGACGTCCA<br>/GTGGAAACGACCT<br>GGACTACATGGAC<br>T | A | G | Gln | Gln | SILENT-CODING | glucoamylase  | Human Gene TREMBL NEW-ID:G2826521 MALTASE-GLUCOAMYLASE (EC 3.2.1.20)-HOMO SAPIENS (HUMAN), 1857 aa.                            | 7.40E-199 | 17<br>(17q25.2<br>) |
| 33 | cg43299024 | 2021 | TGAACGAGCCCTC<br>CAACTCATCAG[G/<br>A]GGCTCTGAGGA<br>CGGCTGCCCAAC<br>A | G | A | Arg | Arg | SILENT-CODING | glucoamylase  | Human Gene TREMBL NEW-ID:G2826521 MALTASE-GLUCOAMYLASE (EC 3.2.1.20)-HOMO SAPIENS (HUMAN), 1857 aa.                            | 7.40E-199 | 17<br>(17q25.2<br>) |
| 34 | cg43969076 | 443  | AATTCCAATGAG<br>CTCTCCAACCAC[G/<br>A]TTTTCTGCCT<br>TTTGATCCAGAC       | G | A | Tyr | Tyr | SILENT-CODING | glucuronidase | Human Gene SWISSPROT-ID:P08236 BETA-GLUCURONIDASE PRECURSOR (EC 3.2.1.31) (BETA-G1) - HOMO SAPIENS (HUMAN), 651 aa.            | 0         | 7<br>(7q21.11<br>)  |
| 35 | cg43969014 | 325  | AATTCCAAGATGAG<br>CTCTCCAACCAC[G/<br>A]TTTTCTGCCT<br>TTTGATCCAGAC     | G | A | Tyr | Tyr | SILENT-CODING | glucuronidase | Human Gene Similar to SWISSPROT-ID:P08236 BETA-GLUCURONIDASE PRECURSOR (EC 3.2.1.31) (BETA-G1) - HOMO SAPIENS (HUMAN), 651 aa. | 7.40E-80  | 5                   |
| 36 | cg43065549 | 880  | GGACCATCTCTGT<br>GACCACACCTGC[G/<br>A]GGACGGCTGTCA<br>GGCCACTACTCGC   | G | A | Ala | Ala | SILENT-CODING | glycoprotein  | Human Gene SWISSPROT-ID:P16452 ERYTHROCYTE MEMBRANE PROTEIN BAND 4.2 (P4.2) (PALLIDIN) - HOMO SAPIENS (HUMAN), 690 aa.         | 0         | 15<br>(15q15<br>)   |

|    |            |      |  |   |   |     |     |               |              |  |           |               |
|----|------------|------|--|---|---|-----|-----|---------------|--------------|--|-----------|---------------|
| 37 | cg43065549 | 991  | ACCCCTGGAAATAG<br>AGAGGATGCTGTT<br>/GTTTCTGAAGAA<br>TGAGGCTCAGC<br>A | T | G | Val | Val | SILENT-CODING | glycoprotein | Human Gene SWISSPROT-ID:P16452 ERYTHROCYTE MEMBRANE PROTEIN BAND 4.2 (P4.2) (PALLIDIN) - HOMO SAPIENS (HUMAN), 690 aa.   | 0         | 15<br>(15q.5) |
| 38 | cg44004239 | 1141 | TGACGTCAATCCAT<br>GTCCAATGTC[CA[C/<br>T]ACCATGCC<br>CCAAAATGCTCT     | C | T | Val | Val | SILENT-CODING | glycoprotein | Human Gene SWISSPROT-ID:Q12889 OVIDUCT-SPECIFIC GLYCOPROTEIN PRECURSOR (OVIDUCTAL GLYCOPROTEIN) (OVIDUCTIN) (ESTROGEN-DEPENDENT OVIDUCT PROTEIN) - HOMO SAPIENS (HUMAN), 678 aa. | 0         |               |
| 39 | cg44004239 | 1846 | GAAGGGATAAAC<br>TGAAGCAATAAA[C/<br>T]TTTTCACGGTGTG<br>GCAAATGTGGACA  | C | T | Lys | Lys | SILENT-CODING | glycoprotein | Human Gene SWISSPROT-ID:Q12889 OVIDUCT-SPECIFIC GLYCOPROTEIN PRECURSOR (OVIDUCTAL GLYCOPROTEIN) (OVIDUCTIN) (ESTROGEN-DEPENDENT OVIDUCT PROTEIN) - HOMO SAPIENS (HUMAN), 678 aa. | 0         |               |
| 40 | cg43957605 | 1677 | AGGACTGTTTCA<br>TTCACTTCAG[A/C<br>]GTGATTCCCATGG<br>GCTCTTCGTGA      | A | C | Thr | Thr | SILENT-CODING | glycoprotein | Human Gene SWISSPROT-ID:Q00013 55 KD ERYTHROCYTE MEMBRANE PROTEIN (P55) - HOMO SAPIENS (HUMAN), 466 aa.  | 3.10E-249 | X (Xq28)      |

|    |            |      |  |   |   |     |               |              |   |           |          |
|----|------------|------|--|---|---|-----|---------------|--------------|---|-----------|----------|
| 41 | cg40915005 | 1229 | ATGTCCTCAGGATTCTACCCAAAGCC[C/T]GTGTGGTGTGAATGGATGGGGTG | C | T | Pro | SILENT-CODING | glycoprotein | Human Gene SWISSNEW-ID:P06126 T-CELL SURFACE GLYCOPROTEIN CD1A PRECURSOR (CD1A ANTIGEN) (T-CELL SURFACE ANTIGEN T6/LEU-6) (HTA1 THYMOCYTE ANTIGEN) - HOMO SAPIENS (HUMAN), 327 aa.[pcds:SWISSPROT-ID:P06126 T-CELL SURFACE GLYCOPROTEIN CD1A PRECURSOR (CD1A ANTIGEN) (T-CELL SURFACE ANTIGEN T6/LEU-6) (HTA1 THYMOCYTE ANTIGEN) - HOMO SAPIENS (HUMAN), 327 aa.] | 2.00E-183 | 1 (1q21) |
| 42 | cg40356255 | 1210 | TGGCAATAATAGTGCCTCCCTGGCTC[T]CTTTGCTATGCCATTATGGT      | C | T | Leu | SILENT-CODING | glycoprotein | Human Gene SWISSNEW-ID:P29016 T-CELL SURFACE GLYCOPROTEIN CD1B PRECURSOR (CD1B ANTIGEN) - HOMO SAPIENS (HUMAN), 333 aa.[pcds:SWISSPROT-ID:P29016 T-CELL SURFACE GLYCOPROTEIN CD1B PRECURSOR (CD1B ANTIGEN) - HOMO SAPIENS (HUMAN), 333 aa.]   | 6.70E-183 | 1 (1q21) |

|    |            |      |   |   |   |     |               |              |   |                       |
|----|------------|------|---|---|---|-----|---------------|--------------|---|-----------------------|
| 43 | cg44004667 | 1183 | CTGTGATATCTACA<br>TCTGGGGGCCCT<br>TGCGCGGACTG<br>TGGGTCTCTCT  | C | T | Leu | SILENT-CODING | glycoprotein | Human Gene Homologous to SWISSNEW-ID:P01732 T-CELL SURFACE GLYCOPROTEIN CD8 ALPHA CHAIN PRECURSOR (T-LYMPHOCYTE DIFFERENTIATION ANTIGEN T8/LEU-2) - HOMO SAPIENS (HUMAN), 235 aa. pcels:SWISSPROT-ID:P01732 T-CELL SURFACE GLYCOPROTEIN CD8 ALPHA CHAIN PRECURSOR (T-LYMPHOCYTE DIFFERENTIATION ANTIGEN T8/LEU-2) - HOMO SAPIENS (HUMAN), 235 aa. | 7.60E-127             |
| 44 | cg43068999 | 544  | AGGGTCTGCAGA<br>GGGTACTTTGTG<br>AAGAGCTAGCCC<br>AAGATTGCTGG   | G | A | Val | SILENT-CODING | glycoprotein | Human Gene Homologous to SWISSPROT-ID:P02743 SERUM AMYLOID P-COMPONENT PRECURSOR (SAP) (9.58 ALPHA-1-GLYCOPROTEIN) - HOMO SAPIENS (HUMAN), 223 aa.  | 1.60E-119             |
| 45 | cg41568631 | 1242 | ATGGCCAGTGGCTG<br>GGCTTGTGGC/<br>A/GGACCAACCA<br>GTGCTGGCTGCC | C | A | Gly | SILENT-CODING | glycoprotein | Human Gene Similar to SWISSPROT-ID:P16452 ERYTHROCYTE MEMBRANE PROTEIN BAND 4.2 (P4.2) (PALLDIN) - HOMO SAPIENS (HUMAN), 690 aa.  | 9.90E-70<br>(14q11.2) |

|    |             |      |   |   |   |     |     |               |              |   |           |                     |
|----|-------------|------|---|---|---|-----|-----|---------------|--------------|---|-----------|---------------------|
| 46 | cg41568631  | 1545 | GCTCTGGAGTC<br>CATCAAGAATGGIC<br>/GCTGGGCTACATG<br>AAGTACGACACGC    | C | G | Gly | Gly | SILENT-CODING | glycoprotein | Human Gene Similar to<br>SWISSPROT-ID:P16452<br>ERYTHROCYTE MEMBRANE<br>PROTEIN BAND 4.2 (P4.2)<br>(PALLIDIN) - HOMO SAPIENS<br>(HUMAN), 690 aa.            | 9.90E-70  | 14<br>(14q11.2<br>) |
| 47 | cg41603916  | 361  | GCATCCAGTGGGT<br>AGGGGACCCCTCGIC<br>/TTGGAAGGATGG<br>CTCCATTGTACATC | C | T | Arg | Arg | SILENT-CODING | glycoprotein | Human Gene Similar to<br>SPTREMBL-ID:Q9J406 IP1=CNS<br>MYELIN P0-LIKE<br>GLYCOPROTEIN - UNKNOWN,<br>202 aa.   | 3.00E-52  | 1 (1q22)            |
| 48 | cg41603916  | 409  | TACACAACCTAGA<br>CTACAGTGACAAIT<br>/CIGGCACGTTCACT<br>TGTGACGTCAAAA | T | C | Asn | Asn | SILENT-CODING | glycoprotein | Human Gene Similar to<br>SPTREMBL-ID:Q9J406 IP1=CNS<br>MYELIN P0-LIKE<br>GLYCOPROTEIN - UNKNOWN,<br>202 aa.   | 3.00E-52  | 1 (1q22)            |
| 49 | cg434317662 | 465  | AGTCCTCTCCGT<br>GGCACCTACGCC[G/<br>CTATGGTTTGAG<br>AAGCCCTCTGCCA    | G | C | Ala | Ala | SILENT-CODING | helicase     | Human Gene Homologous to<br>SWISSPROT-ID:Q12099<br>PROBABLE ATP-DEPENDENT<br>RNA HELICASE FAL1 -<br>SACCHAROMYCES<br>CEREVISIAE (BAKER'S<br>YEAST), 399 aa. | 3.60E-120 | 17                  |
| 50 | cg43983917  | 1353 | AGTCTTACTTTGCC<br>ATTAACCAAA[<br>T]CCCCGACGCCAAG<br>GACTTGAGCAGC    | C | T | Asn | Asn | SILENT-CODING | homeobox     | Human Gene SWISSPROT-<br>ID:P50458 HOMEobox<br>PROTEIN LH-2 - HOMO<br>SAPIENS (HUMAN), 423 aa.  | 4.30E-216 |                     |

|    |            |      |  |   |   |     |     |               |           |   |           |          |
|----|------------|------|--|---|---|-----|-----|---------------|-----------|---|-----------|----------|
| 51 | cg43983917 | 1359 | ACTTTGCCATTAAAC<br>CACAACCCCGAIC/<br>TGGCCAAGGACTTG<br>AAGCAGCTCGCGC     | C | T | Asp | Asp | SILENT-CODING | homeobox  | Human Gene SWISSPROT-ID:P50458 HOMEobox PROTEIN LH-2 - HOMO SAPIENS (HUMAN), 423 aa.  | 4.30E-216 |          |
| 52 | cg42730678 | 979  | TGGAGCGAGCGTG<br>GATCCAGTTTCGC[G<br>T]GGGGGGTTGTT<br>GGGTCAAGTTGCT       | G | T | Ala | Ala | SILENT-CODING | homeobox  | Human Gene SWISSPROT-ID:P28356 HOMEobox PROTEIN HOX-D9 (HOX-4C) (HOX-5.2) - HOMO SAPIENS (HUMAN), 342 aa.   | 2.60E-188 | 2        |
| 53 | cg42714160 | 689  | GTTACCAAGACGCT<br>GGAGGCTGGAGAA[<br>G/A]GAGTTCTACT<br>ACAATCGTACCT<br>GA | G | A | Lys | Lys | SILENT-CODING | homeobox  | Human Gene Homologous to SWISSPROT-ID:P17509 HOMEobox PROTEIN HOX-B6 (HOX-2B) (HOX-2.2) (HU-2) - HOMO SAPIENS (HUMAN), 224 aa.                                  | 1.10E-123 |          |
| 54 | cg43959084 | 810  | TCAGGTAGCGATT<br>GTAGTGAATAATCT/T<br>CTTCTCCAGCTCC<br>AGGGTCGGTAGC       | T | C | Lys | Lys | SILENT-CODING | homeobox  | Human Gene Homologous to SWISSPROT-ID:P09629 HOMEobox PROTEIN HOX-B7 (HOX-2C) (HHO.C1) - HOMO SAPIENS (HUMAN), 217 aa.  | 1.30E-113 |          |
| 55 | cg42359655 | 1124 | GGGAAGGCAATTGC<br>CAATCAGTCCAG[A<br>/G]GGCGGAAGGGAA<br>TGCTCTCCAGCAGG    | A | G | Arg | Arg | SILENT-CODING | hydrolase | Human Gene SWISSPROT-ID:P09848 LACTASE-PHLORIZIN HYDROLASE PRECURSOR (EC 3.2.1.108) (EC 3.2.1.62) (LACTASE-GLYCOSYLCERAMIDASE) - HOMO SAPIENS (HUMAN), 1927 aa. | 0         | 2 (2q21) |

|    |            |      |   |   |   |     |     |               |                |  |           |            |
|----|------------|------|---|---|---|-----|-----|---------------|----------------|--|-----------|------------|
| 56 | cg42359655 | 2468 | ACAGCCAGCGGTT<br>TGGCCTGCACCAIC<br>/TIGICAACTTCAAGC<br>GACAGCAGCAAGT    | C | T | His | His | SILENT-CODING | hydrolase      | Human Gene SWISSPROT-ID:P0948 LACTASE-PHLORIZIN HYDROLASE PRECURSOR (EC 3.2.1.108) (EC 3.2.1.62) (LACTASE-GLYCOSYLCERAMIDASE) - HOMO SAPIENS (HUMAN), 1927 aa. | 0         | 2 (2q21)   |
| 57 | cg42359655 | 4340 | ATCTGGTCACCTGT<br>CAGAACCTGGGC/C/<br>TGTGTCCCCACTAC<br>CGTTTTTCATCT     | C | T | Gly | Gly | SILENT-CODING | hydrolase      | Human Gene SWISSPROT-ID:P0948 LACTASE-PHLORIZIN HYDROLASE PRECURSOR (EC 3.2.1.108) (EC 3.2.1.62) (LACTASE-GLYCOSYLCERAMIDASE) - HOMO SAPIENS (HUMAN), 1927 aa. | 0         | 2 (2q21)   |
| 58 | cg43998672 | 1329 | TGGTGTGGGCCCT<br>GGTGAACCTCTAGIC<br>/A/JACGGCGGCTAAAT<br>GTCTCCCTGGTTGG | C | A | Val | Val | SILENT-CODING | hydroxysteroid | Human Gene SPTRMBL-ID:Q13194 11-BETA-A-HYDROXYSTEROID DEHYDROGENASE TYPE 2 - HOMO SAPIENS (HUMAN), 405 aa.   | 2.00E-220 | 16 (16q22) |
| 59 | cg43922672 | 1689 | GGAAGGTGACTCC<br>AGAGGCCATGCCIC<br>/TIGACCTCAACTCC<br>TCCACTGACTCTG     | C | T | Pro | Pro | SILENT-CODING | interleukin    | Human Gene TREMBL/NEW-ID:G2114410 INTERLEUKIN-16 - HOMO SAPIENS (HUMAN), 631 aa.   | 0         | 15         |

|    |            |     |  |   |   |     |     |               |                   |   |           |          |
|----|------------|-----|--|---|---|-----|-----|---------------|-------------------|---|-----------|----------|
| 60 | cg42908571 | 630 | GTAGTGA <sup>G</sup> GAACA<br>AGCCAGAGCTGTG<br>/CICAGATGAGTAC<br>AAAAGTCTGATC<br>C | G | C | Val | Val | SILENT-CODING | Interleukin       | Human Gene Homologous to SWISSPROT-ID:P05231<br>INTERLEUKIN-6 PRECURSOR<br>(IL-6) (B-CELL STIMULATORY<br>FACTOR 2) (BSF-2)<br>(INTERFERON BETA-2)<br>(HYBRIDOMA GROWTH<br>FACTOR) - HOMO SAPIENS<br>(HUMAN), 212 aa.  | 3.40E-108 | 7 (7p21) |
| 61 | cg43942050 | 181 | AGTGGAA <sup>C</sup> TGAA<br>TGGATCGAGCA[<br>/TTCAGTGACCTGT<br>GCCTTTGAGGACC       | C | T | His | His | SILENT-CODING | interleukinrecept | Human Gene SWISSNEW-ID:P16871 INTERLEUKIN-7<br>RECEPTOR ALPHA CHAIN<br>PRECURSOR (IL-7R-ALPHA)<br>(CDW127) (CD127 ANTIGEN)-<br>HOMO SAPIENS (HUMAN), 459<br>aa.;pels:SWISSPROT-ID:P16871<br>INTERLEUKIN-7 RECEPTOR<br>ALPHA CHAIN PRECURSOR<br>(IL-7R-ALPHA) (CDW127)<br>(CD127 ANTIGEN) - HOMO<br>SAPIENS (HUMAN), 459 aa. | 3.10E-249 | 5 (5p13) |

|    |            |      |   |   |   |     |     |               |        |   |   |    |
|----|------------|------|---|---|---|-----|-----|---------------|--------|---|---|----|
| 62 | cg43145505 | 1249 | TAATATTCGAGA<br>CATTGACAAAGATIC<br>TTATGTTGCAACA<br>GGTATCTACCATG | C | T | Ile | Ile | SILENT-CODING | kinase | Human Gene SWISSNEW-ID:P42336<br>PHOSPHATIDYLINOSITOL 3-KINASE CATALYTIC SUBUNIT, ALPHA ISOFORM (EC 2.7.1.137) (PI3-KINASE P110 SUBUNIT ALPHA) (PTDINS-3-KINASE P110) (P13K) - HOMO SAPIENS (HUMAN), 1068 aa. pct:SWISSPROT-ID:P42336 | 0 | 3  |
| 63 | cg3918241  | 1693 | AGATCTTGAGGA<br>AGGGGAATCTGAIC<br>T]GATGAGTTGAC<br>ATGGATGAGAAC   | C | T | Asp | Asp | SILENT-CODING | kinase | Human Gene SPTRMBL-ID:Q63533 SNF1-RELATED KINASE - RATTUS NORVEGICUS (RAT), 746 aa.   | 0 | 3  |
| 64 | cg4309090  | 1438 | TTCTGACCCACAT<br>GTTTTGTACATTG<br>TICAGACCAAGGA<br>AAACCTTTTG     | C | T | Phe | Phe | SILENT-CODING | kinase | Human Gene SWISSPROT-ID:Q04759 PROTEIN KINASE C, THETA TYPE (EC 2.7.1.-) (NPKC-THETA) - HOMO SAPIENS (HUMAN), 706 aa.   | 0 | 10 |

|    |            |      |   |   |   |     |               |        |   |           |                 |
|----|------------|------|---|---|---|-----|---------------|--------|---|-----------|-----------------|
| 65 | cg43969763 | 2339 | TTAGTATCATTCA<br>TGATCTAA[A/<br>GICCTGAATAATAC<br>CTCTTTGTAACC          | A | G | Lys | SILENT-CODING | kinase | Human Gene SWISSPROT-ID:Q13627 SERINE/THREONINE-SPECIFIC PROTEIN KINASE MINIBRAIN HOMOLOG (EC 2.7.1.-) (HP86) (DYRK) - HOMO SAPIENS (HUMAN), 763 aa.  | 0         | 21<br>(21q22.1) |
| 66 | cg42879455 | 2062 | AGGTATACCAT<br>CATGTACAGTTGT[V<br>CTGGCATGAGAAA<br>GCAGATGAGCGTC        | T | C | Cys | SILENT-CODING | kinase | Human Gene SWISSPROT-ID:Q06187 TYROSINE-PROTEIN KINASE BTK (EC 2.7.1.112) (BRUTON'S TYROSINE KINASE) (AGAMMAGLOBULINAEMIA TYROSINE KINASE) (ATK) (B CELL PROGENITOR KINASE) (BPK) - HOMO SAPIENS (HUMAN), 659 aa. | 0         | X<br>(Xq21.3)   |
| 67 | cg42659872 | 1744 | TGGCTCCGGCTAC<br>ACCAACATCATG[A/<br>C]GGGTGCTAAGC<br>ATATCCTGAGACG<br>C | A | C | Arg | SILENT-CODING | kinase | Human Gene SPTRHEMBL-ID:Q16715 PYRUVATE KINASE (EC 2.7.1.40) - HOMO SAPIENS (HUMAN), 587 aa (fragment).   | 9.80E-308 | 1 (1q21)        |

|    |            |      |   |                      |               |        |  |                       |               |
|----|------------|------|---|----------------------|---------------|--------|--|-----------------------|---------------|
| 68 | cg42506800 | 1323 | GCTTGCCAATTCT<br>CGTCTGTATGCCAC<br>[AAGTACTTCAAG<br>GAGATCTGAATC    | A<br>C<br>Ala<br>Ala | SILENT-CODING | kinase | Human Gene SWISSPROT-ID:Q16654 [PYRUVATE DEHYDROGENASE(LIPOAMID E) KINASE ISOZYME 4 PRECURSOR (EC 2.7.1.99) (PYRUVATE DEHYDROGENASE KINASE ISOFORM 4) - HOMO SAPIENS (HUMAN), 411 aa.] pcls:SPTREMBL-ID:Q16654 PYRUVATE DEHYDROGENASE KINASE ISOFORM 4 - HOMO SAPIENS (HUMAN), 411 aa. | 1.60E-220<br>(7q21.3) | 7             |
| 69 | cg43966621 | 526  | CTGTGGAGTACAT<br>GTAGCTGAAGAGIC<br>TTCGCTCAATCTTC<br>CTCAAGGGAAACAC | C<br>T<br>Arg        | SILENT-CODING | kinase | Human Gene SWISSPROT-ID:Q15119 [PYRUVATE DEHYDROGENASE(LIPOAMID E) KINASE ISOZYME 2 PRECURSOR (EC 2.7.1.99) (PYRUVATE DEHYDROGENASE KINASE ISOFORM 2) - HOMO SAPIENS (HUMAN), 407 aa.] pcls:SPTREMBL-ID:Q15119 PYRUVATE DEHYDROGENASE KINASE - HOMO SAPIENS (HUMAN), 407 aa.           | 3.80E-219<br>17       | 17            |
| 70 | cg43917871 | 1448 | ACATCATATTGGC<br>GCTGGCTGACGGGC<br>TGTACTGCCCT<br>GGCATGCTAGATG     | C<br>T<br>T          | SILENT-CODING | kinase | Human Gene SWISSPROT-ID:PI9138 CASEIN KINASE II, ALPHA CHAIN (CK II) (EC 2.7.1.37) - HOMO SAPIENS (HUMAN), AND BOS TAURUS (BOVINE), 391 aa.  | 2.00E-215<br>(20p13)  | 11<br>(20p13) |

|    |            |      |  |   |   |     |     |               |                |  |           |               |
|----|------------|------|--|---|---|-----|-----|---------------|----------------|--|-----------|---------------|
| 71 | cg43917871 | 1526 | CAGTGTAGAAATA<br>GGGGTGCTCCATT/<br>GIGCCTCTCTTGCA<br>GTAAGCCGTGACT     | T | G | Ala | Ala | SILENT-CODING | kinase         | Human Gene SWISSPROT-ID:P19138 CASEIN KINASE II, ALPHA CHAIN (CK II) (EC 2.7.1.37) - HOMO SAPIENS (HUMAN), AND BOS TAURUS (BOVINE), 391 aa.  | 2.00E-215 | 11<br>(20p13) |
| 72 | cg44131752 | 912  | AGCTCAAATGGTGG<br>CTCTCGTGTGCTC[G/<br>ATCCCCGAAGTGAC<br>CTGCCCTGGTTCGG | G | A | Ser | Ser | SILENT-CODING | kinase         | Human Gene SWISSPROT-ID:Q15599 TYROSINE KINASE ACTIVATOR PROTEIN 1 (TKA-1) - HOMO SAPIENS (HUMAN), 450 aa.   | 7.80E-173 | 16            |
| 73 | cg43969473 | 1765 | AATTCAACCCACT<br>CATCTATGGCAAT/<br>C]GATGTGGATTCT<br>GIGGATGTGCAA      | T | C | Asn | Asn | SILENT-CODING | kinase         | Human Gene SWISSPROT-ID:Q27467 SIMILARITY TO TYROSINE-PROTEIN KINASE-CAENORHABDITIS ELEGANS, 1280 aa.  | 2.10E-154 | 11            |
| 74 | cg44025829 | 610  | AGACCCGGCGTC<br>CCCTGGCCAAGCT<br>/CIGTGGAGTGCTG<br>CCAAGGGGACTGG       | T | C | Ala | Ala | SILENT-CODING | kinasereceptor | Human Gene SWISSPROT-ID:Q04771 ACTIVIN RECEPTOR TYPE I PRECURSOR (EC 2.7.1.-) (ACTR-I) (SERINE/THREONINE-PROTEIN KINASE RECEPTOR RI) (SKR1) (ACTIVIN RECEPTOR-LIKE KINASE 2) (ALK-2) (TGF-B SUPERFAMILY RECEPTOR TYPE I) (TSR-I) - HOMO SAPIENS (HUMAN), 509 aa. | 7.90E-283 | 2             |

|    |            |      |  |   |   |     |               |     |   |           |               |
|----|------------|------|--|---|---|-----|---------------|-----|---|-----------|---------------|
| 75 | cg43318277 | 1107 | CTCACGGCTTGCAG<br>TCATCTGGTCCIG/A<br>CCCTAGCACTCCCT<br>CCTCTCCTCGGC      | G | A | Pro | SILENT-CODING | MHC | Human Gene SPTREMBL-ID:Q02646 MHC BINDING PROTEIN 2 - HOMO SAPIENS (HUMAN), 2500 aa.  | 1.20E-247 | 6             |
| 76 | cg43966144 | 632  | TTAACACGGGGA<br>GCCTGTGATGCTG/<br>A GCCCTGCTATGTG<br>TGGGGCTCTATC        | G | A | Leu | SILENT-CODING | MHC | Human Gene Homologous to SWISSPROT-ID:P28068 CLASS II HISTOCOMPATIBILITY ANTIGEN, M BETA CHAIN PRECURSOR - HOMO SAPIENS (HUMAN), 263 aa.                      | 9.10E-147 | 6<br>(6p21.3) |
| 77 | cg42686658 | 644  | CCCCCTGTGATCAAT<br>ATCACCTGGCTTA/<br>GICGCAACGGCCA<br>AACTGTCACTGAG<br>G | A | G | Leu | SILENT-CODING | MHC | Human Gene Homologous to SWISSPROT-ID:P06340 HLA CLASS II HISTOCOMPATIBILITY ANTIGEN, DZ ALPHA CHAIN PRECURSOR (MHC DN-ALPHA) - HOMO SAPIENS (HUMAN), 250 aa. | 3.70E-134 | 6<br>(6p21.3) |
| 78 | cg42686658 | 857  | CACCACAGATGC<br>CATGGAGACCCCTG<br>A GTCCTGGCCCTG<br>GGCCTGGCCATCG        | G | A | Leu | SILENT-CODING | MHC | Human Gene Homologous to SWISSPROT-ID:P06340 HLA CLASS II HISTOCOMPATIBILITY ANTIGEN, DZ ALPHA CHAIN PRECURSOR (MHC DN-ALPHA) - HOMO SAPIENS (HUMAN), 250 aa. | 3.70E-134 | 6<br>(6p21.3) |
| 79 | cg42686658 | 869  | CCATGGAGACCCCT<br>GGTCTGTGCCCTG/<br>A GGCCTGGCCATC<br>GGCCTGGGGCT        | G | A | Leu | SILENT-CODING | MHC | Human Gene Homologous to SWISSPROT-ID:P06340 HLA CLASS II HISTOCOMPATIBILITY ANTIGEN, DZ ALPHA CHAIN PRECURSOR (MHC DN-ALPHA) - HOMO SAPIENS (HUMAN), 250 aa. | 3.70E-134 | 6<br>(6p21.3) |

|    |            |     |   |   |   |     |     |               |     |  |           |               |
|----|------------|-----|---|---|---|-----|-----|---------------|-----|--|-----------|---------------|
| 80 | cg42686658 | 881 | TGGCTCTGCCCCCTG<br>GGCCCTGGCCAT[CT<br> GGCCCTGGGGCT<br>TCCTCGTGGGCA       | C | T | Ile | Ile | SILENT-CODING | MHC | Human Gene Homologous to<br>SWISSPROT-ID:P06340 HLA<br>CLASS II<br>HISTOCOMPATIBILITY<br>ANTIGEN, DZ ALPHA CHAIN<br>PRECURSOR (MHC DN-ALPHA)<br>- HOMO SAPIENS (HUMAN),<br>250 aa. | 3.70E-134 | 6<br>(6p21.3) |
| 81 | cg42686658 | 893 | TGGGCCCTGGCCAT<br>CGGCCCTGGGGIC<br>/GTTCCCTCGTGGGC<br>ACCGTCCATCA         | C | G | Gly | Gly | SILENT-CODING | MHC | Human Gene Homologous to<br>SWISSPROT-ID:P06340 HLA<br>CLASS II<br>HISTOCOMPATIBILITY<br>ANTIGEN, DZ ALPHA CHAIN<br>PRECURSOR (MHC DN-ALPHA)<br>- HOMO SAPIENS (HUMAN),<br>250 aa. | 3.70E-134 | 6<br>(6p21.3) |
| 82 | cg42686658 | 905 | TCGGGCCCTGGTGGG<br>CTTCCTCGTGGG[C/<br>T]ACCGTCTCATC<br>ATCATGGGCACAT      | C | T | Gly | Gly | SILENT-CODING | MHC | Human Gene Homologous to<br>SWISSPROT-ID:P06340 HLA<br>CLASS II<br>HISTOCOMPATIBILITY<br>ANTIGEN, DZ ALPHA CHAIN<br>PRECURSOR (MHC DN-ALPHA)<br>- HOMO SAPIENS (HUMAN),<br>250 aa. | 3.70E-134 | 6<br>(6p21.3) |
| 83 | cg38337333 | 279 | GTTCCTCATTA[G/C<br>CCTGTGACCC[AT<br>]GCACACGGAGGG<br>ACCTACAGATGTC        | A | T | Pro | Pro | SILENT-CODING | MHC | Human Gene Homologous to<br>SPTREMBL-ID:Q93368 HLA<br>CLASS I INHIBITORY NK<br>RECEPTOR - HOMO SAPIENS<br>(HUMAN), 455 aa.   | 1.80E-113 | 19            |
| 84 | cg38337333 | 492 | TTGACATCTTACCAT<br>CTATCCAGGGAG/[G/<br>A]GGGAAGCCCA<br>TGAACTTAGGCTC<br>C | G | A | Glu | Glu | SILENT-CODING | MHC | Human Gene Homologous to<br>SPTREMBL-ID:Q93368 HLA<br>CLASS I INHIBITORY NK<br>RECEPTOR - HOMO SAPIENS<br>(HUMAN), 455 aa.   | 1.80E-113 | 19            |

|    |            |     |  |   |   |     |               |              |  |           |          |
|----|------------|-----|--|---|---|-----|---------------|--------------|--|-----------|----------|
| 85 | cg38337333 | 699 | CTCTTAGTAGTGG<br>CCTTCACCCACTA<br>[GAACCAAGCTCA<br>AAACTGGTATCG          | T | A | Thr | SILENT-CODING | MHC          | Human Gene Homologous to<br>SPTREMBL-ID:Q93368 HLA<br>CLASS I INHIBITORY NK<br>RECEPTOR - HOMO SAPIENS<br>(HUMAN), 455 aa. | 1.80E-113 | 19       |
| 86 | cg38337333 | 774 | GGTACTCAAGTGGC<br>CATCATCCTCTTC/<br>TTACCATCCTCCC<br>TTCTTTCTCCCTC       | C | T | Phe | SILENT-CODING | MHC          | Human Gene Homologous to<br>SPTREMBL-ID:Q93368 HLA<br>CLASS I INHIBITORY NK<br>RECEPTOR - HOMO SAPIENS<br>(HUMAN), 455 aa. | 1.80E-113 | 19       |
| 87 | cg38337333 | 783 | TGGCCCATCATCCTC<br>TTACCATCCTT/C<br>JCCCTTCTTCTCT<br>TCATGCTGGT          | T | C | Leu | SILENT-CODING | MHC          | Human Gene Homologous to<br>SPTREMBL-ID:Q93368 HLA<br>CLASS I INHIBITORY NK<br>RECEPTOR - HOMO SAPIENS<br>(HUMAN), 455 aa. | 1.80E-113 | 19       |
| 88 | cg43984759 | 649 | AGGAGCTCAAGCG<br>TGAGGCCGAGACIC<br>/T]CTACGGGAGCG<br>GGAAGGGAGGAG<br>T   | C | T | Thr | SILENT-CODING | misc_channel | Human Gene SPTREMBL-<br>ID:Q14193 H-DRK1 K(+)<br>CHANNEL - HOMO SAPIENS<br>(HUMAN), 858 aa.                                | 0         | 20       |
| 89 | cg39660131 | 990 | TCATGGGCAACCT<br>AAGGCCACAAGTGT<br>C/TGTGGCGCAACTT<br>CACAGCGCTCAAC<br>G | C | T | Cys | SILENT-CODING | misc_channel | Human Gene SPTREMBL-<br>ID:Q14524 SODIUM CHANNEL<br>ALPHA SUBUNIT - HOMO<br>SAPIENS (HUMAN), 2016 aa.                      | 0         | 3 (3p24) |

|    |            |      |  |   |   |     |     |               |              |  |           |                     |
|----|------------|------|--|---|---|-----|-----|---------------|--------------|--|-----------|---------------------|
| 90 | cg44963814 | 717  | CGGAATACTCTGGC<br>CATCACCTCTGAA[A/<br>G]AGCAAGAGAGAA<br>CTGCACGGGCCTC<br>C | A | G | Glu | Glu | SILENT-CODING | misc_channel | Human Gene Homologous to<br>SWISSPROT-ID:Q07699<br>SODIUM CHANNEL BETA-1<br>SUBUNIT PRECURSOR - HOMO<br>SAPIENS (HUMAN), 218<br>aa.[pels:TREMBL NEW.<br>ID:G2804300 VOLTAGE-GATED<br>SODIUM CHANNEL BETA-1<br>SUBUNIT - HOMO SAPIENS<br>(HUMAN), 218 aa. | 2.20E-113 | 19<br>(19q13.1<br>) |
| 91 | cg21413267 | 870  | AGAGTGGCGAGTG<br>GGTCATCTGGAA[C<br>T]GCCGTGGCAC<br>CTACAAACCCAGG<br>A      | C | T | Asp | Asp | SILENT-CODING | misc_channel | Human Gene Similar to<br>SPTREMBL-ID:P91197 SIMILAR<br>TO LIGAND-GATED IONIC<br>CHANNEL PROTEIN -<br>CAENORHABDITIS ELEGANS,<br>461 aa.  | 7.90E-79  |                     |
| 92 | cg21413267 | 909  | ACAACACCAAGGAA<br>GTACGAGGTGCTGC<br>[T]GCCGAGATCTA<br>CCGGGACATCACC<br>T   | C | T | Cys | Cys | SILENT-CODING | misc_channel | Human Gene Similar to<br>SPTREMBL-ID:P91197 SIMILAR<br>TO LIGAND-GATED IONIC<br>CHANNEL PROTEIN -<br>CAENORHABDITIS ELEGANS,<br>461 aa.  | 7.90E-79  |                     |
| 93 | cg3000465  | 1160 | AGAGGGCTTCTTG<br>CAGAAACTTCC[A/<br>C]AAATTACTTGC<br>ATGAAAAGATCATG         | A | C | Pro | Pro | SILENT-CODING | misc_channel | Human Gene Similar to<br>SPTREMBL-ID:P91197 SIMILAR<br>TO LIGAND-GATED IONIC<br>CHANNEL PROTEIN -<br>CAENORHABDITIS ELEGANS,<br>461 aa.  | 6.10E-70  | 8<br>(8p11.2)       |

|    |            |      |   |   |   |     |               |             |   |           |               |
|----|------------|------|---|---|---|-----|---------------|-------------|---|-----------|---------------|
| 94 | cg30421838 | 3766 | GTCTAGGATGGAG<br>ATCCTACAAACA[C<br>T]GTCAGGGCA<br>GATGCTGTATTTG         | C | T | His | SILENT-CODING | nucl_recept | Human Gene SWISSNEW-ID:P06401 PROGESTERONE RECEPTOR (PR) - HOMO SAPIENS (HUMAN), 933 aa. polis:SWISSPROT-ID:P06401 PROGESTERONE RECEPTOR (PR) - HOMO SAPIENS (HUMAN), 933 aa. | 0         | 11<br>(11q22) |
| 95 | cg30421838 | 4114 | ATAACTTGCATGA<br>TCTTGTCAAACA[A/<br>G]CTTCATCTGTAC<br>TGCTTGAATACAT     | A | G | Gln | SILENT-CODING | nucl_recept | Human Gene SWISSNEW-ID:P06401 PROGESTERONE RECEPTOR (PR) - HOMO SAPIENS (HUMAN), 933 aa. polis:SWISSPROT-ID:P06401 PROGESTERONE RECEPTOR (PR) - HOMO SAPIENS (HUMAN), 933 aa. | 0         | 11<br>(11q22) |
| 96 | cg43947341 | 713  | TTAGTCCGCCAAA<br>TCCCCAGGGCAC[A<br>T]GTTGCCACGAA<br>CTTCAGTACGGGA<br>T  | A | G | Asn | SILENT-CODING | nuclease    | Human Gene Homologous to SWISSPROT-ID:P07992 DNA EXCISION REPAIR PROTEIN ERCC-1 - HOMO SAPIENS (HUMAN), 297 aa.   | 1.10E-115 |               |
| 97 | cg43939230 | 4226 | TCCCTGTGACCCA<br>GGCAGGGTGCATGA<br>[G]GTGACACTGGT<br>CGTGACCTGGCCA<br>G | A | G | Thr | SILENT-CODING | oncogene    | Human Gene SPTREMBL-ID:Q99907 LATENT TRANSFORMING GROWTH FACTOR-BETA-BINDING PROTEIN-2 - HOMO SAPIENS (HUMAN), 1821 aa.   | 0         | 14<br>(14q4)  |

|     |            |      |   |   |   |     |     |               |               |   |                     |          |
|-----|------------|------|---|---|---|-----|-----|---------------|---------------|---|---------------------|----------|
| 98  | cg42674136 | 1447 | CGGCACACAGGCC<br>GCTGCCGGAGCIC<br>TGTGGCCACCC<br>CAGCCCCCTGGCCA     | C | T | Ala | Ala | SILENT-CODING | oncogene      | Human Gene SWISSPROT-ID:P31314 HOMEOBOX PROTEIN HOX-11 (TCL-3 PROTO-ONCOGENE) - HOMO SAPIENS (HUMAN), 330 aa.                                     | 3.70E-182           | 10       |
| 99  | cg41972699 | 742  | AGAAACTCGGGT<br>CTCCCACATACATIC/<br>TATCAAACTCGCTG<br>CCCAACGCCCGTT | C | T | Ile | Ile | SILENT-CODING | oncogene      | Human Gene Similar to SWISSPROT-ID:Q64010 PROTO-ONCOGENE C-CRK (P38) (ADAPTER MOLECULE CRK)- MUS MUSCULUS (MOUSE), 304 aa.                        | 2.40E-84<br>(22q11) | 22       |
| 100 | cg42849556 | 963  | CTGCAACTACCTTG<br>AACCAGTGAAGC/<br>TTCGGGATCCACC<br>CTCAGGAGGAGCC   | C | T | Leu | Leu | SILENT-CODING | oxidase       | Human Gene SWISSPROT-ID:P19878 NEUTROPHIL CYTOSOL FACTOR 2 (NCF-2) (NEUTROPHIL NADPH OXIDASE FACTOR 2) (P67-PHOX) - HOMO SAPIENS (HUMAN), 526 aa. | 2.80E-287           | 1 (1q25) |
| 101 | cg43996195 | 1310 | CAGCATGACCTGG<br>CACTGTACTTCG[G/<br>A]GAAAGTGG<br>GATTTCACCGTAGT    | G | A | Pro | Pro | SILENT-CODING | phosphorylase | Human Gene SWISSPROT-ID:P00491 PURINE NUCLEOSIDE PHOSPHORYLASE (EC 2.4.2.1) (INOSINE PHOSPHORYLASE) (PNP) - HOMO SAPIENS (HUMAN), 289 aa.         | 2.40E-155           |          |

|     |            |      |   |   |   |     |               |     |               |               |   |           |   |
|-----|------------|------|---|---|---|-----|---------------|-----|---------------|---------------|---|-----------|---|
| 102 | cg43996195 | 1421 | TTGCAACTTGTAGG<br>TCGGTGCTTAGT[G/<br>A]TGAGACAGAAAG<br>CCATTCTGCAGTGT | G | A | His | SILENT-CODING | His | SILENT-CODING | phosphorylase | Human Gene SWISSPROT-ID:P00491 PURINE NUCLEOSIDE PHOSPHORYLASE (EC 2.4.2.1) (INOSINE PHOSPHORYLASE) (PNP) - HOMO SAPIENS (HUMAN), 289 aa.   | 2.40E-155 |   |
| 103 | cg43948227 | 372  | TTTACAGTTTCTT<br>ACTGGCATCATCIA[T<br>A]ATGTCAGAAATCT<br>GTTCCCTCAGCT  | A | T | Ile | SILENT-CODING |     |               | polymerase    | Human Gene Similar to SWISSNEW-ID:P33999 ACTIVATED RNA POLYMERASE II TRANSCRIPTIONAL COACTIVATOR P15 (PC4) (P14) - HOMO SAPIENS (HUMAN), 126 aa.[pcls:SWISSPROT-ID:P33999 ACTIVATED RNA POLYMERASE II TRANSCRIPTIONAL COACTIVATOR P15 (PC4) (P14) - HOMO SAPIENS (HUMAN), 126 aa. | 5.40E-62  | 5 |



|     |            |      |   |   |   |     |     |               |                    |   |           |                 |
|-----|------------|------|---|---|---|-----|-----|---------------|--------------------|---|-----------|-----------------|
| 105 | cg43051431 | 1683 | TCAACACCTGTCCTG<br>ACCCGGAGGAAIT<br>CJGGGTCTTACGAA<br>GTTGACTACAACA | T | C | Asp | Asp | SILENT-CODING | potassium_chan nel | Human Gene SWISSPROT-ID:P48051 G PROTEIN- ACTIVATED INWARD RECTIFIER POTASSIUM CHANNEL 2 (GIRK2) (POTASSIUM CHANNEL, INWARDLY RECTIFYING, SUBFAMILY J, MEMBER 6) (KATP-2) (BIR1) (KIR3.2)- HOMO SAPIENS (HUMAN), 423 aa. pcls:TREMBLNEW. ID:G1518526 INWARDLY RECTIFYING POTASSIUM CHANNEL KIR3.2 - HOMO SAPIENS (HUMAN), 423 aa. | 1.60E-227 | 16              |
| 106 | cg43920929 | 1081 | GCAGGGATCACCTG<br>CACCCCTCTGGGIC<br>GJACCATGAIGCTC<br>ATCCAGCTGTCTA | C | G | Va  | Val | SILENT-CODING | proteaseinhib      | Human Gene SWISSPROT-ID:P07093 GLIA DERIVED NEXIN PRECURSOR (GDN) (PROTEASE NEXIN 1) (PN-1) (PROTEASE INHIBITOR 7)- HOMO SAPIENS (HUMAN), 398 aa.   | 1.20E-208 | 2               |
| 107 | cg43059041 | 624  | AGTCAGAACCCAG<br>CTTAGAAATGACIC<br>TATGGGCAATGC<br>CTTGTTCTTGATG    | C | T | Thr | Thr | SILENT-CODING | proteaseinhib      | Human Gene Similar to SWISSPROT-ID:P17475 ALPHA-1-ANTIPROTEINASE PRECURSOR (ALPHA-1-ANTITRYPsin) (ALPHA-1-PROTEINASE INHIBITOR)- RATTUS NORVEGICUS (RAT), 411 aa.   | 4.40E-83  | 14<br>(14q2.1 ) |

|     |            |      |  |             |                   |                             |   |           |               |
|-----|------------|------|--|-------------|-------------------|-----------------------------|---|-----------|---------------|
| 108 | cg40148056 | 1385 | GGAGGACAGGGCAA<br>CTCATCACCGAAC<br>/TTAGTCAATCAGCA<br>AGATGAACCAGCT          | C<br>T<br>A | Leu<br>Leu<br>Glu | SILENT-<br>CODING<br>struct | Human Gene SPTREMBL-<br>ID:Q9277 SYNAPSIN IIB -<br>HOMO SAPIENS (HUMAN), 478<br>aa.   | 2.90E-260 | 3 (3p)        |
| 109 | cg42894986 | 1002 | ACCCGTTCTCTGC<br>CCACCCACTGAA[G/<br>A]GCCCGAGACCGT<br>GACCTCTGGGG            | G<br>A      | Glu<br>Glu        | SILENT-<br>CODING<br>struct | Human Gene SPTREMBL-<br>ID:Q28686 50-KDA<br>DYSTROPHIN-ASSOCIATED<br>GLYCOPROTEIN PRECURSOR -<br>ORYCTOLAGUS CUNICULUS<br>(RABBIT), 387 aa.   | 1.40E-180 | 17            |
| 110 | cg43961212 | 2160 | TCTGGAAGCCCCA<br>CATCCCTCTGAGCA/<br>GIAGTCGACTGATC<br>CGCTGGGAACCA           | A<br>G      | Leu<br>Leu        | SILENT-<br>CODING<br>struct | Human Gene Homologous to<br>TREMBL/NEW-ID:GI703715<br>PANTOPHYSIN=SYNAPTOPHY<br>SIN HOMOLOG - MUS SP, 261<br>aa.  | 2.40E-114 | 7             |
| 111 | cg42898003 | 497  | TCATCAGAGATTG<br>GATCTCCCTCGTC[C/<br>A]GTCACTGTGCTCC<br>CCGGAGGGCCCTGA       | C<br>A      | Thr<br>Thr        | SILENT-<br>CODING<br>struct | Human Gene Similar to<br>SWISSPROT-ID:P02585<br>TROPONIN C, SKELETAL<br>MUSCLE - HOMO SAPIENS<br>(HUMAN), 159 aa.   | 1.50E-80  | 20<br>(20q12) |
| 112 | cg43960684 | 788  | GCTTTGAGGGAGGA<br>GGCGCGGGTTGGCGC[G/<br>G]GACGACACTGA<br>GGCGGGCCATCCGC<br>G | C<br>G      | Arg<br>Arg        | SILENT-<br>CODING<br>struct | Human Gene Similar to<br>SWISSPROT-ID:P02535<br>KERATIN, TYPE I<br>CYTOSKELETAL 10<br>(CYTOKERATIN 10) (56 KD<br>CYTOKERATIN) (KERATIN,<br>TYPE I CYTOSKELETAL 59 KD)<br>MUS MUSCULUS (MOUSE),<br>569 aa. | 8.30E-58  | 8             |

|     |            |      |   |                       |                                 |   |  |   |          |    |
|-----|------------|------|---|-----------------------|---------------------------------|---|--|---|----------|----|
| 113 | cg43958714 | 1049 | TTCGGAAAGGGCA<br>AGCAGTGAACCTG<br>TCATGATGATGATGC<br>CACCAATATGCCA<br>G | G<br>C<br>C<br>C<br>G | Leu<br>Leu<br>Leu<br>Leu<br>Leu | SILENT-CODING<br>SILENT-CODING<br>SILENT-CODING<br>SILENT-CODING<br>SILENT-CODING | synthase<br>synthase<br>synthase<br>synthase<br>synthase | Human Gene Similar to<br>SPTREMBL-ID:Q42761<br>SQUALENE SYNTHASE (EC<br>2.5.1.21) (FARNESYL-<br>DIPHOSPHATE<br>FARNESYLTRANSFERASE)<br>(FARNESYLTRANSFERASE)<br>(PRESQUALENE-DI-<br>DIPHOSPHATE<br>SYNTHASE) - GLYCYYRRHIZA<br>GLABRA, 412 aa.  | 9.20E-83 | 8  |
| 114 | cg43124627 | 901  | ACACCCACAGCGAG<br>TTTGGTTAGGTA/<br>TTTATCTGAAAT<br>GGAAGGTCTGGC         | A<br>T<br>Gly<br>Gly  | T<br>Gly<br>Gly<br>Gly          | SILENT-CODING<br>SILENT-CODING<br>SILENT-CODING<br>SILENT-CODING                  | synthase<br>synthase<br>synthase<br>synthase             | Human Gene Similar to<br>SWISSNEW-ID:P39062<br>ACETYL-COENZYME A<br>SYNTHETASE (EC 6.2.1.1)<br>(ACETATE--COA LIGASE)<br>(ACYL- ACTIVATING<br>ENZYME) (ACETYL-COA<br>SYNTHASE) - BACILLUS<br>SUBTILIS, 572<br>aa.<br>lpcis:SWISSPROT-ID:P39062<br>ACETYL-COENZYME A<br>SYNTHETASE (EC 6.2.1.1)<br>(ACETATE--COA LIGASE)<br>(ACYL- ACTIVATING<br>ENZYME) (ACETYL-COA<br>SYNTHASE) - BACILLUS<br>SUBTILIS, 572 aa. | 7.70E-79 | 16 |

|     |            |     |  |   |   |     |     |               |          |  |          |
|-----|------------|-----|--|---|---|-----|-----|---------------|----------|--|----------|
| 115 | cg43968419 | 906 | TCTTCTCCAAACAGT<br>CTGCCACCCGCA[T<br>CTCGTGTGGCTGCG<br>CCTCCAAGGCC | A | T | Ala | Ala | SILENT-CODING | synthase | Human Gene Similar to<br>SWISSNEW-ID:P53556-8-<br>AMINO-7-OXONONANOATE<br>SYNTHASE (EC 2.3.1.47) (7-<br>KETO-8-AMINO- PELARGONIC<br>ACID SYNTHETASE) (7-KAP<br>SYNTHETASE)(L-ALANINE--<br>PIMELYL COA LIGASE)-<br>BACILLUS SUBTILIS, 389<br>aa.lpcis:SWISSPROT-ID:P53556<br>8-AMINO-7-OXONONANOATE<br>SYNTHASE (EC 2.3.1.47) (7-<br>KETO-8-AMINO- PELARGONIC<br>ACID SYNTHETASE) (7-KAP<br>SYNTHETASE)(L-ALANINE--<br>PIMELYL COA LIGASE)-<br>BACILLUS SUBTILIS, 389 aa. | 9.90E-70 |
|     |            |     |  |   |   |     |     |               |          |  |          |
|     |            |     |  |   |   |     |     |               |          |  |          |
|     |            |     |  |   |   |     |     |               |          |  |          |
|     |            |     |  |   |   |     |     |               |          |  |          |

|     |            |      |   |        |     |     |               |          |   |          |
|-----|------------|------|---|--------|-----|-----|---------------|----------|---|----------|
| 116 | cgt3064068 | 1484 | TGTTGGTCTGGCC<br>TCGCAGTTCCTG/A<br>TCCCCATGACCCAG<br>AACAGCTACCA        | G<br>A | Leu | Leu | SILENT-CODING | synthase | Human Gene Similar to<br>SWISSNEW-ID:P39062<br>ACETYL-COENZYME A<br>SYNTHETASE (EC 6.2.1.1)<br>(ACETATE--COA LIGASE)<br>(ACYL- ACTIVATING<br>ENZYME) (ACETYL-COA<br>SYNTHASE) - BACILLUS<br>SUBTILIS, 572<br>aa.lcids;SWISSPROT-ID:P39062<br>ACETYL-COENZYME A<br>SYNTHETASE (EC 6.2.1.1)<br>(ACETATE--COA LIGASE)<br>(ACYL- ACTIVATING<br>ENZYME) (ACETYL-COA<br>SYNTHASE) - BACILLUS<br>SUBTILIS, 572 aa. | 7.40E-65 |
| 117 | cgt3064068 | 1622 | TCACAGGGAAAT<br>TCAACGGGCCAAAG<br>[A]CTTCGAGACAA<br>GGAGTGGAAAGATG<br>T | G<br>A | Lys | Lys | SILENT-CODING | synthase | Human Gene Similar to<br>SWISSNEW-ID:P39062<br>ACETYL-COENZYME A<br>SYNTHETASE (EC 6.2.1.1)<br>(ACETATE--COA LIGASE)<br>(ACYL- ACTIVATING<br>ENZYME) (ACETYL-COA<br>SYNTHASE) - BACILLUS<br>SUBTILIS, 572<br>aa.lcids;SWISSPROT-ID:P39062<br>ACETYL-COENZYME A<br>SYNTHETASE (EC 6.2.1.1)<br>(ACETATE--COA LIGASE)<br>(ACYL- ACTIVATING<br>ENZYME) (ACETYL-COA<br>SYNTHASE) - BACILLUS<br>SUBTILIS, 572 aa. | 7.40E-65 |

|     |            |      |  |   |   |     |               |     |   |           |    |
|-----|------------|------|--|---|---|-----|---------------|-----|---|-----------|----|
| 118 | cg41084924 | 1278 | TGACTCTCCCCGAC<br>CCGTCCCACCA[CT]<br>JGGTCTCCACAGCA<br>CTCCCGACAGGCC   | C | T | His | SILENT-CODING | tm7 | Human Gene SWISSPROT-ID:P14416 D(2) DOPAMINE RECEPTOR - HOMO SAPIENS (HUMAN), 443 aa.                 | 1.70E-241 | 11 |
| 119 | cg41084924 | 1662 | TCGCAAGGCCCT<br>CCTGAAGATCCT[C/<br>T]CACTGCTGACTC<br>TGCTGCCCTGCCG     | C | T | Leu | SILENT-CODING | tm7 | Human Gene SWISSPROT-ID:P14416 D(2) DOPAMINE RECEPTOR - HOMO SAPIENS (HUMAN), 443 aa.                 | 1.70E-241 | 11 |
| 120 | cg41084924 | 606  | TCCTCGTGCACACA<br>CTGGTCATGCC[C/A]<br>TGGGGTTGCTACC<br>TGGAGGTGGTAG    | C | A | Pro | SILENT-CODING | tm7 | Human Gene SWISSPROT-ID:P14416 D(2) DOPAMINE RECEPTOR - HOMO SAPIENS (HUMAN), 443 aa.                 | 1.70E-241 | 11 |
| 121 | cg43985000 | 1471 | TTGCTCTTGTGG<br>TTCCCTCTCA[C/T]<br>TTAAGCCGTATT<br>GAAGAAAATCG         | C | T | His | SILENT-CODING | tm7 | Human Gene SWISSPROT-ID:P25101 ENDOTHELIN-1 RECEPTOR PRECURSOR (ET-A) - HOMO SAPIENS (HUMAN), 427 aa. | 1.60E-236 | 4  |
| 122 | cg43985000 | 1507 | TATTGAAGAAAC<br>TGTGTATAACG[A/<br>G]ATGGACAAAGAA<br>CCGATGTGAATTA<br>C | A | G | Glu | SILENT-CODING | tm7 | Human Gene SWISSPROT-ID:P25101 ENDOTHELIN-1 RECEPTOR PRECURSOR (ET-A) - HOMO SAPIENS (HUMAN), 427 aa. | 1.60E-236 | 4  |

|     |            |      |  |   |   |     |               |     |  |           |   |
|-----|------------|------|--|---|---|-----|---------------|-----|--|-----------|---|
| 123 | cg44930578 | 561  | ACGTGAAACACCGA<br>CATCTACTCCAAAG/<br>A/GTGCTGGTGACC<br>GCCGTTAACCTGG | G | A | Lys | SILENT-CODING | tm7 | Human Gene SWISSPROT-ID:P30989 NEUROTENSIN (HIGH-AFFINITY LEVOCABASTINE-SENSITIVE NEUROTENSIN RECEPTOR) (NTRH) - HOMO SAPIENS (HUMAN), 418 aa.   | 5.00E-217 |   |
| 124 | cg3003519  | 1263 | ATTCCCTCATTTGCT<br>AGGACCCCTTA/C/T<br>AAAAAGCACCTGA<br>ACATACCTACTG  | C | T | Tyr | SILENT-CODING | tm7 | Human Gene SWISSNEW-ID:P32247 BOMBESIN RECEPTOR SUBTYPE-3 (BRS-3) - HOMO SAPIENS (HUMAN), 399 aa.[pcl:SWISSPROT-ID:P32247 BOMBESIN RECEPTOR SUBTYPE-3 (BRS-3) - HOMO SAPIENS (HUMAN), 399 aa.[pcl:TREMBLNEW-ID:E1240254 BOMBESIN RECEPTOR SUBTYPE-3 (UTERINE BOMBESIN RECEPTOR, BRS-3) - HOMO SAPIENS (HUMAN), 399 aa. | 3.00E-212 | X |

|     |            |      |  |   |   |     |     |               |     |  |           |                     |
|-----|------------|------|--|---|---|-----|-----|---------------|-----|--|-----------|---------------------|
| 125 | cg3003519  | 711  | CTTATGCTGTGATC<br>ATTCAAGTGGG[C/T]<br>[ATCCCTTGAAATG<br>CTATTCTCATCA     | C | T | Gly | Gly | SILENT-CODING | tm7 | Human Gene SWISSPROT-<br>ID:P32247 BOMBESIN<br>RECEPTOR SUBTYPE-3 (BRS-3)<br>- HOMO SAPIENS (HUMAN),<br>399 aa.[pds:SWISSPROT-<br>ID:P32247 BOMBESIN<br>RECEPTOR SUBTYPE-3 (BRS-3)<br>- HOMO SAPIENS (HUMAN),<br>399 aa.[pcls:TREMBLNEW-<br>ID:E1240254 BOMBESIN<br>RECEPTOR SUBTYPE-3<br>(UTERINE BOMBESIN<br>RECEPTOR, BRS-3) - HOMO<br>SAPIENS (HUMAN), 399 aa. | 3.00E-212 | X                   |
| 126 | cg43969010 | 1182 | TCCGAAAGAAGTC<br>TTGGGAGGTGTA[TC<br>[T]CAGGGAGTGTG<br>CCAGAAAAGGGGC<br>T | C | T | Tyr | Tyr | SILENT-CODING | tm7 | Human Gene SWISSPROT-<br>ID:P30411 B2 BRADYKININ<br>RECEPTOR (BK-2 RECEPTOR)-<br>HOMO SAPIENS (HUMAN), 391<br>aa.  | 9.00E-211 | 12<br>(14q22.1<br>) |
| 127 | cg43263108 | 1097 | AGACACCCCTTCC<br>CAGCTCGCTC[CA<br>JGGAGGGAGGAC<br>CCAAGGGCCCCT<br>T      | C | A | Ser | Ser | SILENT-CODING | tm7 | Human Gene SWISSPROT-<br>ID:P43119 PROSTACYCLIN<br>RECEPTOR (PROSTANOID IP<br>RECEPTOR) (PGI RECEPTOR)-<br>HOMO SAPIENS (HUMAN), 386<br>aa.  | 8.30E-208 | 19<br>(19q13.3<br>) |
| 128 | cg43263108 | 272  | GCCCCCTGGGCCCTC<br>GGGT[GCTGGT[C/<br>G]ACCGGACTGGCG<br>GCCACCGACCTGC     | C | G | Val | Val | SILENT-CODING | tm7 | Human Gene SWISSPROT-<br>ID:P43119 PROSTACYCLIN<br>RECEPTOR (PROSTANOID IP<br>RECEPTOR) (PGI RECEPTOR)-<br>HOMO SAPIENS (HUMAN), 386<br>aa.  | 8.30E-208 | 19<br>(19q13.3<br>) |

|     |            |      |  |   |   |     |     |               |     |   |           |               |
|-----|------------|------|--|---|---|-----|-----|---------------|-----|---|-----------|---------------|
| 129 | cg43267238 | 1220 | CCAGACTGGTCCCT<br>GGTGGTGGTGGCIA<br>/G/GCTTCGTGCGTC<br>TGCTGGACTCCCCA  | A | G | Ala | Ala | SILENT-CODING | tm7 | Human Gene SWISSPROT-ID:P41145 KAPPA-TYPE OPIOID RECEPTOR (KOR-1) - HOMO SAPIENS (HUMAN), 380 aa. | 2.10E-204 | 8<br>(8q11.2) |
| 130 | cg43267238 | 392  | CAGCACTACCAT<br>GGAATCCCCGATC<br>/T]CAGATTTCCGC<br>GGGGAGCCGGGCC       | C | T | Ile | Ile | SILENT-CODING | tm7 | Human Gene SWISSPROT-ID:P41145 KAPPA-TYPE OPIOID RECEPTOR (KOR-1) - HOMO SAPIENS (HUMAN), 380 aa. | 2.10E-204 | 8<br>(8q11.2) |
| 131 | cg43267238 | 413  | CGATCCACATCTTC<br>CGGGGGAGCCIG<br>T]GGCCCTACCTGC<br>GCCCGAGCGCT        | G | T | Pro | Pro | SILENT-CODING | tm7 | Human Gene SWISSPROT-ID:P41145 KAPPA-TYPE OPIOID RECEPTOR (KOR-1) - HOMO SAPIENS (HUMAN), 380 aa. | 2.10E-204 | 8<br>(8q11.2) |
| 132 | cg43264978 | 155  | TGGATCTGCACCTC<br>TTGACTACTCA/C<br>IGAGCCAGGGAAC<br>TTCTCGGACATCA      | A | C | Ser | Ser | SILENT-CODING | tm7 | Human Gene TREMBLINEW-ID:G2736282 G PROTEIN COUPLED RECEPTOR - HOMO SAPIENS (HUMAN), 362 aa.      | 1.40E-196 |               |
| 133 | cg3001696  | 1154 | CGCTGCACCTGTG<br>CATCGCGCTGGGIC<br>/TTACGCCAATAG<br>CAGCCTCAACCCC<br>G | C | T | Gly | Gly | SILENT-CODING | tm7 | Human Gene SWISSPROT-ID:P41143 DELTA-TYPE OPIOID RECEPTOR (DOR-1) - HOMO SAPIENS (HUMAN), 372 aa. | 2.10E-195 | 1<br>(1p36.1) |

|     |            |     |  |   |   |     |     |               |     |   |                            |
|-----|------------|-----|--|---|---|-----|-----|---------------|-----|---|----------------------------|
| 134 | cg3001696  | 815 | TGGCTGTGACCCG<br>TCCCCGGGACGGIG<br>/TGGAGTGTTG<br>CATGCTCCAGTCCC     | G | T | Gly | Gly | SILENT-CODING | tm7 | Human Gene SWISSPROT-ID:P41143 DELTA-TYPE OPIOID RECEPTOR (DOR-1) - HOMO SAPIENS (HUMAN), 372 aa.   | 2.10E-195<br>1<br>(1p36.1) |
| 135 | cg42704646 | 407 | TGGCCTTCCGATC<br>ACCATGCTGCT[C/G<br>]ACTGGTTCTGTGG<br>GAAACGCACTGG   | C | G | Leu | Leu | SILENT-CODING | tm7 | Human Gene SWISSPROT-ID:P43115 PROSTAGLANDIN E2 RECEPTOR, EP3 SUBTYPE (PROSTANOID EP3 RECEPTOR) (PGE RECEPTOR, EP3 SUBTYPE) - HOMO SAPIENS (HUMAN), 390 aa. | 3.10E-194<br>1<br>(1p31.2) |
| 136 | cg43326635 | 347 | GGGATGCCACCT<br>CTGGCTCATCGTC/<br>GTCGCTGGCGGTG<br>GCTGATGGGCCG      | C | G | Val | Val | SILENT-CODING | tm7 | Human Gene SWISSPROT-ID:P30542 ADENOSINE A1 RECEPTOR - HOMO SAPIENS (HUMAN), 326 aa.  | 1.10E-173<br>1             |
| 137 | cg3003708  | 358 | CCATTCCTCTCTGT<br>GGCTGTCTCAC[A/<br>]GCAAGATGTATTIC<br>GTTTTCATGTTCG | A | G | Thr | Thr | SILENT-CODING | tm7 | Human Gene TREMBL NEW-ID:E1246031 OLFACTORY RECEPTOR - HOMO SAPIENS (HUMAN), 312 aa.  | 2.50E-160                  |
| 138 | cg3003708  | 787 | GGTGGAAAGCCCT<br>CTCCACCTGGGT/<br>CTCTCACCTGGCT<br>GTGGTTCTCCCTCT    | T | C | Gly | Gly | SILENT-CODING | tm7 | Human Gene TREMBL NEW-ID:E1246031 OLFACTORY RECEPTOR - HOMO SAPIENS (HUMAN), 312 aa.  | 2.50E-160                  |

|     |            |     |  |   |   |     |     |                   |     |  |           |
|-----|------------|-----|--|---|---|-----|-----|-------------------|-----|--|-----------|
| 139 | cg3003708  | 841 | ACAGGCCATTCAAT<br>TGCTGTGTTATTTT/<br>CIAACCCTCTGCCC<br>TCCCACCTAGCTG   | T | C | Phe | Phe | SILENT-<br>CODING | tm7 | Human Gene TREMBL NEW-<br>ID:EL246031 OLFACTORY<br>RECEPTOR - HOMO SAPIENS<br>(HUMAN), 312 aa.   | 2.50E-160 |
| 140 | cg36729339 | 537 | ACTCTCCAATGTAC<br>TTTTTCCTCTC[C/T]<br>AACCTCTCTCTCTT<br>GGACCTCTGCT    | C | T | Ser | Ser | SILENT-<br>CODING | tm7 | Human Gene SWISSPROT-<br>ID:Q15062 OLFACTORY<br>RECEPTOR-LIKE PROTEIN<br>FAT11 -HOMO SAPIENS<br>(HUMAN), 316 aa.   | 1.90E-153 |
| 141 | cg38841806 | 717 | GACATCAAGGCCA<br>CGGTGCCAACCTC<br>/GICGCCATCTGCA<br>GCCAAAGAAGAAG<br>T | C | G | Leu | Leu | SILENT-<br>CODING | tm7 | Human Gene Similar to<br>SWISSPROT-ID:P30975<br>TACHYKININ-LIKE PEPTIDES<br>RECEPTOR 99D (DTKR) -<br>DROSOPHILA<br>MELANOOGASTER (FRUIT FLY),<br>519 aa. | 2.10E-67  |
| 142 | cg38841806 | 723 | AGGGCACCGTGC<br>CAACCTCCGCAAT/<br>CJCTGCAGGCCAAG<br>AGAAAGTTGTGA       | T | C | His | His | SILENT-<br>CODING | tm7 | Human Gene Similar to<br>SWISSPROT-ID:P30975<br>TACHYKININ-LIKE PEPTIDES<br>RECEPTOR 99D (DTKR) -<br>DROSOPHILA<br>MELANOOGASTER (FRUIT FLY),<br>519 aa. | 2.10E-67  |
| 143 | cg38841806 | 96  | CAGCCTTCCTCCATG<br>CCCCAGCTGGCA[G/<br>A]CTGGCACTGTGG<br>GCACCAAGCTACC  | G | A | Gln | Gln | SILENT-<br>CODING | tm7 | Human Gene Similar to<br>SWISSPROT-ID:P30975<br>TACHYKININ-LIKE PEPTIDES<br>RECEPTOR 99D (DTKR) -<br>DROSOPHILA<br>MELANOOGASTER (FRUIT FLY),<br>519 aa. | 2.10E-67  |

|     |            |      |  |   |   |     |               |                  |  |           |          |
|-----|------------|------|--|---|---|-----|---------------|------------------|--|-----------|----------|
| 144 | cg43040273 | 1966 | CCCTGTGCTGATCTG<br>GTCATGGGCCCTG/<br>AIGCAGTGGTGC<br>TTGGGGCGCCC         | G | A | Leu | SILENT-CODING | tm7              | Human Gene Similar to<br>SWISSPROT-ID:Q24563<br>DOPAMINE RECEPTOR 2 -<br>DROSOPHILA<br>MELANOOGASTER (FRUIT FLY),<br>539 aa.   | 2.00E-58  | 5 (5q32) |
| 145 | cg43040273 | 2237 | CTTGCCCCATTCA<br>TGACTGGTAC[C/<br>A]GGGCCACCCACC<br>AGGAAGCCATCAA        | C | A | Arg | SILENT-CODING | tm7              | Human Gene Similar to<br>SWISSPROT-ID:Q24563<br>DOPAMINE RECEPTOR 2 -<br>DROSOPHILA<br>MELANOOGASTER (FRUIT FLY),<br>539 aa.   | 2.00E-58  | 5 (5q32) |
| 146 | cg43336100 | 687  | TTGAAAGCGTCCA<br>TCCAGTGAGCCJA<br>/TJATGAGGCTTGA<br>GTCTTTTAGTGCCT       | A | T | Pro | SILENT-CODING | tsf              | Human Gene SWISSPROT-<br>ID:P26022 PENTAXIN-<br>RELATED PROTEIN PTX3<br>PRECURSOR (TUMOR<br>NECROSIS FACTOR-<br>INDUCIBLE PROTEIN TSG-14)-<br>HOMO SAPIENS (HUMAN), 381<br>aa.   | 2.20E-207 | 3 (3q25) |
| 147 | cg21646034 | 376  | GTTGTGAGCAGAGA<br>TGCCAGAACCAA[A<br>/G]GTGGACCGAAC<br>ACCATTACATATG<br>G | A | G | Lys | SILENT-CODING | transcriptfactor | Human Gene SWISSPROT-<br>ID:Q06545 GA BINDING<br>PROTEIN BETA-2 CHAIN<br>(GABP-BETA-2 SUBUNIT)<br>(TRANSCRIPTION FACTOR<br>E4IF1-47) (GAPBP2) - HOMO<br>SAPIENS (HUMAN), 347 aa. | 9.00E-179 | 15       |

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|-----|------------|------|---|---|---|-----|-----|---------------|-------------|---|-----------|----------|
| 148 | cg43916882 | 1608 | TGGCAGCTTACCAAG<br>CACACTGCCTCCIA/<br>GICCGTCAATAAAG<br>GCACTGAAGGTCT | A | G | Gly | Gly | SILENT-CODING | transferase | Human Gene SWISSPROT-ID:P39656 DOLICHYL-DIPHOSPHOOLIGOSACCHARIDE-PROTEIN GLYCOSYLTRANSFERASE 48 KD SUBUNIT PRECURSOR (EC 2.4.1.19) (OLIGOSACCHARYL TRANSFERASE 48 KD SUBUNIT) (DD03T 48 KD SUBUNIT) (KIAA0115) (HA0643) - HOMO SAPIENS (HUMAN), 456 aa.   | 5.30E-245 | 1        |
| 149 | cg2537639  | 294  | TGGCTCCCATTGTC<br>TGGGAGGGCACIA/<br>GTTCAAACTCGAC<br>ATCCCTCAACGAGC   | A | G | Thr | Thr | SILENT-CODING | transferase | Human Gene SWISSPROT-ID:P16442 FUCOSYLGlycoprotein ALPHA-N-acetylgalactosaminyltransferase (EC 2.4.1.40) (histo-blood group A transferase) (A-transferase) / FUCOSYLGlycoprotein 3-alpha-galactosyltransferase (EC 2.4.1.37) (histo-blood group B transferase) (B-transferase) (NAGAT)- HOMO SAPIENS (HUMAN), 354 aa. | 6.50E-192 | 9 (9q34) |

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|-----|-----------|-----|--|---|---|-----|-----|---------------|-------------|---|-----------|----------|
| 150 | cg2537639 | 654 | ACGTGGACATGGAGTCGGGACCA[CT]GTGGCGTGGAGATCCTGACTCGC | C | T | His | His | SILENT-CODING | transferase | Human Gene SWISSPROT-ID:P16442<br>FUCOSYLGlycoprotein<br>ALPHA-N-ACETYLGLALACTOSAMINYL TRANSFERASE (EC 2.4.1.40)<br>(HISTO-BLOOD GROUP A TRANSFERASE) (A TRANSFERASE) /<br>FUCOSYLGlycoprotein 3-<br>ALPHA-GALACTOSYLTRANSFERASE<br>(EC 2.4.1.37) (HISTO-BLOOD<br>GROUP B TRANSFERASE) (B<br>TRANSFERASE) (NAGAT)-<br>HOMO SAPIENS (HUMAN), 354 aa. | 6.50E-192 | 9 (9q34) |
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|-----|-----------|-----|--|---|-----|-----|---------------|-------------|--|-----------|----------|
| 151 | cg2537639 | 678 | ACGTGGCGTGGGA G<br>GATCCCTGACTCCIG/<br>AICGTTCGGCACC<br>CTGCACCCGGCT | A | Pro | Pro | SILENT-CODING | transferase | Human Gene SWISSPROT-ID:P16442<br>FUCOSYLYCOPROTEIN-<br>ALPHA-N-<br>ACETYLGLALACTOSAMINYLT<br>RANSFERASE (EC 2.4.1.40)<br>(HISTO-BLOOD GROUP A<br>TRANSFERASE) (A<br>TRANSFERASE) /<br>FUCOSYLYCOPROTEIN 3-<br>ALPHA-<br>GALACTOSYLTRANSFERASE<br>(EC 2.4.1.37) (HISTO-BLOOD<br>GROUP B TRANSFERASE) (B<br>TRANSFERASE) (NAGAT)-<br>HOMO SAPIENS (HUMAN), 354<br>aa. | 6.50E-192 | 9 (9q34) |
|     |           |     |  |   |     |     |               |             |  |           |          |
|     |           |     |  |   |     |     |               |             |  |           |          |
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|-----|-----------|-----|---|---|---|-----|-----|---------------|-------------|--|-----------|----------|
| 152 | cg2537639 | 768 | GGCCCCAGTCCCA<br>GGCCTACATCC[C/C]<br>TAAAGGACGAGGG<br>CGATTCTACTAAC | C | T | Pro | Pro | SILENT-CODING | transferase | Human Gene SWISSPROF-<br>ID:PI6442<br>FUCOSYLYLCOPROTEIN<br>ALPHA-N- | 6,50E-192 | 9 (9q34) |
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|-----|------------|-----|--|---|---|-----|---------------|-------------|---|-----------|----------|
| 153 | cg2537639  | 927 | ACGAGAGGCCACCT<br>GAACAAGTACCTTG<br>/A/CTGCGCACAA<br>ACCCACCAAGGTG<br>C  | G | A | Leu | SILENT-CODING | transferase | Human Gene SWISSPROT-ID:P16442<br>FUCOSYGLYCOPROTEIN ALPHA-N-ACETYLGLALACTOSAMINYLT<br>RANSFERASE (EC 2.4.1.40)<br>(HISTO-BLOOD GROUP A<br>TRANSFERASE) (A<br>TRANSFERASE) /<br>FUCOSYGLYCOPROTEIN 3-<br>ALPHA-GALACTOSYL TRANSFERASE<br>(EC 2.4.1.37) (HISTO-BLOOD<br>GROUP B TRANSFERASE) (B<br>TRANSFERASE) (NAGAT)-<br>HOMO SAPiens (HUMAN), 354<br>aa. | 6.50E-192 | 9 (9q34) |
| 154 | cg44000740 | 732 | GGGGAGATACTGG<br>CTCACCCAGGAATA<br>/C/ACAGGGAAACAT<br>CACCTTATGCCAC<br>A | A | C | Val | SILENT-CODING | transferase | Human Gene Homologous to<br>SWISSPROT-ID:P30711<br>GLUTATHIONE S-<br>TRANSFERASE THETA 1 (EC<br>2.5.1.18) (CLASS-THETA)-<br>HOMO SAPIENS (HUMAN), 239<br>aa.  | 1.60E-117 | 16       |

|     |            |      |   |   |   |     |               |           |   |           |            |
|-----|------------|------|---|---|---|-----|---------------|-----------|---|-----------|------------|
| 155 | cg38869466 | 1185 | ACGCAGTGGCCGT<br>GGGCTCCCTCTGC/<br>TGGCCTTCGCC<br>AGTCTTCTAGGTT     | C | T | Cys | SILENT-CODING | transport | Human Gene SWISSPROT-ID:P30825 HIGH-AFFINITY CATIONIC AMINO ACID TRANSPORTER-1 (CAT-1) (CAT1) (SYSTEM Y+ BASIC AMINO ACID TRANSPORTER) (ECOTROPIC RETROVIRAL LEUKEMIA RECEPTOR HOMOLOG) (ERR) (ECOTROPIC RETROVIRUS RECEPTOR HOMOLOG) - HOMO SAPIENS (HUMAN), 629 aa. | 0         | 13         |
| 156 | cg40351913 | 1347 | CCATCGCCACGCT<br>CCCTCTGTCTCTCA/<br>GIGCCTGGGCCGTG<br>GTCTTCTTCATCA | A | G | Ser | SILENT-CODING | transport | Human Gene SWISSPROT-ID:Q01959 SODIUM-DEPENDENT DOPAMINE TRANSPORTER (DA TRANSPORTER) (DAT) - HOMO SAPIENS (HUMAN), 620 aa.   | 0         | 5 (5p15.3) |
| 157 | cg43964039 | 1719 | GATGGAACAGCTC<br>CTCGGGTCTCTTG/<br>ATTCACTTGGCTG<br>GCTCCCCCTGCC    | G | A | Asp | SILENT-CODING | transport | Human Gene SWISSPROT-ID:Q11166 GLUCOSE TRANSPORTER TYPE 1, ERYTHROCYTE/BRAIN - HOMO SAPIENS (HUMAN), 492 aa.  | 1.60E-259 | 1          |
| 158 | cg43992017 | 1656 | GCGGCTGGTGGTG<br>GATGGGTTGGCGC[G<br>/G]GGGGTGCAGCC<br>TCCACCCCTCCCC | C | G | Pro | SILENT-CODING | transport | Human Gene SPTREMBL-ID:Q14728 TETRACYCLINE TRANSPORTER-LIKE PROTEIN mRNA - HOMO SAPIENS (HUMAN), 455 aa.  | 4.40E-241 |            |

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|-----|------------|------|--|---|---|-----|-----|---------------|--------------|--|---|----------|
| 159 | cg43948629 | 1238 | CGCCTGTAATGGC<br>TGTGAACATGCTC/<br>TIACCCCAGCAGGAG<br>GTCCCTGTCTGTTA   | C | T | Leu | Leu | SILENT-CODING | UNCLASSIFIED | Human Gene SWISSNEW-<br>ACC:Q15031 PROBABLE<br>LEUCYL-TRNA SYNTHETASE,<br>MITOCHONDRIAL<br>PRECURSOR (EC 6.1.1.4)<br>(LEUCINE--TRNA LIGASE)<br>(LEURS) (KIAA0028) - Homo<br>sapiens (Human), 903 aa. | 0 | 3        |
| 160 | cg43955093 | 2875 | CATTGACTAGGG<br>CTGTGGGGCAT[C<br>/GICGCCAGGT<br>CCCTCCATCAGAG<br>G     | C | G | Arg | Arg | SILENT-CODING | UNCLASSIFIED | Human Gene SPTREMBL-<br>ACC:Q16084 P130 - HOMO<br>SAPIENS (HUMAN), 1139 aa.  | 0 | 16       |
| 161 | cg43955093 | 3385 | AGCAGGCCAAGAG<br>AGATCTTGAA[<br>T]GCATCTGTCC<br>AGAATAACGATA           | C | T | Ala | Ala | SILENT-CODING | UNCLASSIFIED | Human Gene SPTREMBL-<br>ACC:Q16084 P130 - HOMO<br>SAPIENS (HUMAN), 1139 aa.  | 0 | 16       |
| 162 | cg43055918 | 517  | CGCTGGCATAGGA<br>CATGGCGGGCTT[G<br>T]CCCCCGCAGA<br>GCTCTGGGGCTA<br>C   | G | T | Gly | Gly | SILENT-CODING | UNCLASSIFIED | Human Gene SWISSPROT-<br>ACC:P42694 HYPOTHETICAL<br>PROTEIN KIAA0054 - Homo<br>sapiens (Human), 1942 aa.   | 0 | 17       |
| 163 | cg43974592 | 254  | AAATAACAAGGCA<br>TTGAAGAATGGCT<br>[A]GACGGAGCGGA<br>AGACGAAGGAAAG<br>G | T | A | Ala | Ala | SILENT-CODING | UNCLASSIFIED | Human Gene REMIREMBL-<br>ACC:E1296438 SEQUENCE 28<br>FROM PATENT WO97/27323 -<br>UNIDENTIFIED, 1829 aa.  | 0 | 2 (2q34) |

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|-----|------------|------|---|---|---|-----|-----|---------------|--------------|--|---|---------------------|
| 164 | cg43956384 | 206  | AAGGACGGAAACGCC<br>TGCCACCATGGAA[C<br>T]AGTAGCACCTG<br>GAGCCCCAAGACC<br>A | C | T | Asp | Asp | SILENT-CODING | UNCLASSIFIED | Human Gene SWISSPROT-<br>ACC:P13866<br>SODIUM/GLUCOSE<br>COTRANSPORTER 1<br>(NA(+)/GLUCOSE<br>COTRANSPORTER 1) (HIGH<br>AFFINITY SODIUM/GLUCOSE<br>COTRANSPORTER) - Homo<br>sapiens (Human), 664 aa. | 0 | 22<br>(22q13.1<br>) |
| 165 | cg44025634 | 2757 | TGAAAGTATTCAA<br>TCCCAGAAGGAA[A<br>A/G]CTGGAAATTG<br>CCCTTCTGTGTTCTA<br>G | A | G | Lys | Lys | SILENT-CODING | UNCLASSIFIED | Human Gene SWISSNEW-<br>ACC:P00450 CERULOPLASMIN<br>PRECURSOR (EC 1.16.3.1)<br>(FEROXIDASE) - Homo sapiens<br>(Human), 1065 aa.  | 0 | 3 (3q21)            |
| 166 | cg43940037 | 2472 | GCTGGGGCACTGC<br>TAGCCTCAGGGT[T<br>A]GCCAGCACCTC<br>CTCAGCCCCCGCG<br>C    | T | A | Ala | Ala | SILENT-CODING | UNCLASSIFIED | Human Gene SWISSPROT-<br>ACC:P4250 GLYCYL-TRNA<br>SYNTHETASE (EC 6.1.1.14)<br>(GLYCINE--TRNA LIGASE)<br>(GLYRS) - Homo sapiens<br>(Human), 665 aa.   | 0 | 7(7p15)             |
| 167 | cg44024279 | 481  | AAAACCAAGCTTAC<br>TGCCCTTCTGGAA[A<br>G]AACTTGTCCAT<br>GAGAAAAGAAATT       | A | G | Glu | Glu | SILENT-CODING | UNCLASSIFIED | Human Gene SWISSPROT-<br>ACC:P0271 ALPHA-<br>FETOPROTEIN PRECURSOR<br>(ALPHA-FETOGLLOBULIN)<br>(ALPHA-1- FETOPROTEIN)-<br>Homo sapiens (Human), 609 aa.  | 0 |                     |

|     |            |      |   |   |   |     |     |               |              |  |           |          |
|-----|------------|------|---|---|---|-----|-----|---------------|--------------|--|-----------|----------|
| 168 | cg43926814 | 1122 | CATGAGTTTGATC<br>CCAGCTCTCTCT<br>TCCCCGGCTTCT<br>GGGCCATTCTC          | C | T | Glu | Glu | SILENT-CODING | UNCLASSIFIED | Human Gene SWISSNEW-<br>ACC:Q13573 NUCLEAR<br>PROTEIN SKIP (SNW1<br>PROTEIN) (NUCLEAR<br>RECEPTOR COACTIVATOR<br>NCOA-62) - Homo sapiens<br>(Human), 536 aa. | 5.00E-289 | 14       |
| 169 | cg40918088 | 1778 | TGGAGCTGGAAT<br>TACTGTATGATA/<br>GJGCCTTAGCAGCT<br>GCTGATGAGCTT       | A | G | Glu | Glu | SILENT-CODING | UNCLASSIFIED | Human Gene SWISSPROT-<br>ACC:P51854 TRANSKETOLASE<br>2 (EC 2.2.1.1) (TK 2)<br>(TRANSKETOLASE RELATED<br>PROTEIN) - Homo sapiens<br>(Human), 557 aa.          | 1.80E-287 | X (Xq28) |
| 170 | cg43966985 | 1242 | TCAACACCTTACGT<br>CCACTTCCAAGGIG<br>/TAAAGATGAAAGGG<br>CTTCTCCCTGCTGG | G | T | Gly | Gly | SILENT-CODING | UNCLASSIFIED | Human Gene SWISSPROT-<br>ACC:P01019<br>ANGIOTENSINOGEN<br>PRECURSOR - Homo sapiens<br>(Human), 485 aa.   | 3.90E-257 | 1 (1q42) |
| 171 | cg43924009 | 770  | TGGCTTGACAAA<br>TTCCTGAAGACIA<br>TTCGATCCATGTAA<br>GTGGACTGTCTTG      | A | T | Arg | Arg | SILENT-CODING | UNCLASSIFIED | Human Gene SPTREMBL-<br>ACC:O43411 HYPOTHETICAL<br>49.3 KD PROTEIN - HOMO<br>SAPIENS (HUMAN), 442 aa<br>(fragment).  | 6.90E-239 |          |
| 172 | cg42913861 | 2186 | CTGGCAGCTGCC<br>CTCACAGTAGTTC<br>GJCGGTAGTAGCCG<br>GTGGGTGCTATGA      | C | G | Gly | Gly | SILENT-CODING | UNCLASSIFIED | Human Gene SWISSPROT-<br>ACC:P09529 INHIBIN BETA B<br>CHAIN PRECURSOR (ACTIVIN<br>BETA-B CHAIN) - Homo sapiens<br>(Human), 407 aa.                           | 3.00E-227 | 2 (2cen) |

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|-----|------------|------|--|---|---|-----|---------------|----------------|--|-----------|------------------|
| 173 | cg42913861 | 2354 | GCCGAGCCTGCAC<br>CACCACAAAGGGI<br>CT]CGGTGCGACTC<br>TTCGCCTGGTCCA    | C | T | Arg | SILENT-CODING | UNCLASSIFIED D | Human Gene SWISSPROT-<br>ACC:P09529 INHIBIN BETA B<br>CHAIN PRECURSOR (ACTIVIN<br>BETA-B CHAIN) - Homo sapiens<br>(Human), 407 aa.   | 3.00E-227 | 2 (2ccn)         |
| 174 | cg43929685 | 256  | CATAGAAGGCCAG<br>GAGTCAGGAGAC]<br>C/TGGTTCTGTC<br>CTGGATTATACAC<br>C | C | T | Gln | SILENT-CODING | UNCLASSIFIED D | Human Gene SWISSPROT-<br>ACC:P29080 (2'-<br>5')OLIGOADENYLATE<br>SYNTHETASE 1B (EC 2.7.7.-)<br>(2'-5')OLIGO(A) SYNTHETASE<br>1B) (2'-5'A SYNTHETASE 1B) -<br>Mus musculus (Mouse), 414 aa. | 2.40E-225 | 12               |
| 175 | cg43929685 | 268  | GGAGTCAGGGAGAC<br>CTGGGTCTCTGC/C/<br>TTGGATTATAACAC<br>CAGCTCACTGAGG | C | T | Gln | SILENT-CODING | UNCLASSIFIED D | Human Gene SWISSPROT-<br>ACC:P29080 (2'-<br>5')OLIGOADENYLATE<br>SYNTHETASE 1B (EC 2.7.7.-)<br>(2'-5')OLIGO(A) SYNTHETASE<br>1B) (2'-5'A SYNTHETASE 1B) -<br>Mus musculus (Mouse), 414 aa. | 2.40E-225 | 12               |
| 176 | cg43918561 | 53   | CCATGCCACCCC<br>CGACGCCACAC[G<br>C]CCACAGGCCAA<br>GGGCTTCCGCAGG<br>G | G | C | Thr | SILENT-CODING | UNCLASSIFIED D | Human Gene SWISSPROT-<br>ACC:P04177 TYROSINE 3-<br>MONOOXYGENASE (EC<br>1.14.16.2) (TYROSINE 3-<br>HYDROXYLASE) (TH) - Rattus<br>norvegicus (Rat), 498 aa.                                 | 2.10E-224 | 11<br>(11p15.5 ) |

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| 177 | cg42343176 | 1885 | ATTAATGAAATTTC<br>CTGAAGACTGTIA/<br>GIAGAAGTACAAC<br>GAGAAATCCCTTT  | A | G | Val | SILENT-CODING | UNCLASSIFIED | Human Gene SWISSPROT-<br>ACC:P14902 INDOLEAMINE<br>2,3-DIOXYGENASE (EC<br>1.13.11.42) (IDO)<br>(INDOLEAMINE-PYRROLE 2,3-<br>DIOXYGENASE) - Homo sapiens<br>(Human), 403 aa.        | 3.90E-218 | 8 (8p12)      |
| 178 | cg43956382 | 1146 | AAAACAATGATAT<br>CGATGAAGTTTC/<br>TJAATCCCACAGCT<br>CCCTTATAACAAAC  | C | T | Ile | SILENT-CODING | UNCLASSIFIED | Human Gene SPTREMBL-<br>ACC:Q99816 TUMOR<br>SUSCEPTIBILITY PROTEIN -<br>HOMO SAPIENS (HUMAN), 390<br>aa.   | 4.90E-211 | 11            |
| 179 | cg43984681 | 979  | CACCATGAAAGCAG<br>TTGCTGCGGGCC[<br>]TGGAGGAGGGCC<br>GCGTGCGGAAAGT   | C | T | Leu | SILENT-CODING | UNCLASSIFIED | Human Gene SWISSPROT-<br>ACC:O15382 BRANCHED-<br>CHAIN AMINO ACID<br>AMINOTRANSFERASE,<br>MITOCHONDRIAL<br>PRECURSOR (EC 2.6.1.42)<br>(BCAT(M)) - Homo sapiens<br>(Human), 392 aa. | 1.30E-210 | 19<br>(19q13) |
| 180 | cg43984681 | 1074 | TCCGTACAAAGA<br>CAGGAACCTCCAIC<br>[T]ATTCCCCACATG<br>GAAAAATGGGCCTG | C | T | His | SILENT-CODING | UNCLASSIFIED | Human Gene SWISSPROT-<br>ACC:O15382 BRANCHED-<br>CHAIN AMINO ACID<br>AMINOTRANSFERASE,<br>MITOCHONDRIAL<br>PRECURSOR (EC 2.6.1.42)<br>(BCAT(M)) - Homo sapiens<br>(Human), 392 aa. | 1.30E-210 | 19<br>(19q13) |

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|-----|------------|------|---|---|---|-----|---------------|--------------|--|-----------|----|
| 181 | cg43950996 | 1762 | CTGGGGTGGAGAC<br>GTCAGAGCTGCCIA<br>TGIGGGGAGGGGC<br>TCCTGCGCACAG<br>C   | A | G | Pro | SILENT-CODING | UNCLASSIFIED | Human Gene SPTREMBL-<br>ACC:P7345 ESE-IB - HOMO<br>SAPIENS (HUMAN), 371 aa.  | 6.20E-204 | 1  |
| 182 | cg44024506 | 988  | ACCAAGCTGCTCGT<br>AGTACACAGGCAI<br>G/AIGCACATTCTCCCT<br>TGCCCTACCTCCATG | G | A | Leu | SILENT-CODING | UNCLASSIFIED | Human Gene SPTREMBL-<br>ACC:O60704<br>TYROSYLPROTEIN<br>SULFOTRANSFERASE 2 -<br>HOMO SAPIENS (HUMAN), 377<br>aa.                                 | 1.90E-200 | 22 |
| 183 | cg43980381 | 1114 | CTACCGCCAACTA<br>TGACTTTGTCCTC/<br>GIAAGAACGGAC<br>CTTCACCAAGGGA<br>G   | C | G | Leu | SILENT-CODING | UNCLASSIFIED | Human Gene SWISSNEW.<br>ACC:Q03385 GUANINE<br>NUCLEOTIDE DISSOCIATION<br>STIMULATOR RALGDS FORM<br>A (RALGEF) - Mus musculus<br>(Mouse), 852 aa. | 5.60E-191 | 9  |

|     |            |      |   |   |   |     |     |               |                |  |                       |
|-----|------------|------|---|---|---|-----|-----|---------------|----------------|--|-----------------------|
| 184 | cg42650960 | 501  | TCCCCTGGCAGAA<br>CTACCACTGAAIC<br>TGTACTGGATGGA<br>GGAGGAATACCGC<br>C | C | T | Asn | Asn | SILENT-CODING | UNCLASSIFIED D | Human Gene SWISSPROT-<br>ACC:Q10981 GALACTOSIDE 2-L-FUCOSYLTRANSFERASE 2-<br>(EC 2.4.1.69) (GDP-L-FUCOSE-BETA-D-GALACTOSIDE 2-ALPHA-L-FUCOSYLTRANSFERASE 2)<br>(ALPHA(1,2)FT 2)<br>(FUCOSYLTRANSFERASE 2)<br>(SECRETOR BLOOD GROUP ALPHA-2-<br>FUCOSYLTRANSFERASE)<br>(SECRETOR FACTOR) (SE)<br>(SE2) - Homo sapiens (Human),<br>343 aa. | 2.00E-189             |
| 185 | cg43249389 | 1497 | ACATCCAGGGTT<br>GTTCGACGCCGTTC<br>TACCGACATCATC<br>ATGCCAACAAACC      | C | T | Val | Val | SILENT-CODING | UNCLASSIFIED D | Human Gene SWISSPROT-<br>ACC:P09471 GUANINE NUCLEOTIDE-BINDING PROTEIN G(O), ALPHA SUBUNIT 1 - Homo sapiens (Human), 353 aa.   | 1.40E-188             |
| 186 | cg43946951 | 615  | CAGTGACGGCAGG<br>GTCAAAGTCCTTG/<br>A]GGCTAGCCCTCG<br>TTAAGGCTGTAGA    | G | A | Ala | Ala | SILENT-CODING | UNCLASSIFIED D | Human Gene SWISSPROT-<br>ACC:P09467 FRUCTOSE-1,6-BISPHOSPHATASE (EC 3.1.3.11)<br>(D-FRUCTOSE-1,6-BISPHOSPHATE 1-PHOSPHOHYDROLASE)<br>(FBPASE) - Homo sapiens (Human), 337 aa.  | 3.50E-178<br>(9q22.2) |

|     |            |      |  |   |   |     |     |               |              |   |           |                                  |
|-----|------------|------|--|---|---|-----|-----|---------------|--------------|---|-----------|----------------------------------|
| 187 | cg43248117 | 1054 | AACCAGCCCCACTG<br>TGAGAAAGACCACTG<br>G/C]GIGTCAGTC<br>TTTGGGAATGGCA<br>G | G | C | Thr | Thr | SILENT-CODING | UNCLASSIFIED | Human Gene SWISSPROT-<br>ACC:Q14894 MU-CRYSTALLIN   | 1.20E-161 | <sup>16</sup><br>(16p13.1<br>.1) |
| 188 | cg44027049 | 482  | CCACAAATGTTAGG<br>AGGGTATTTTAAC<br>T]ATCCCTCCAGTT<br>AACAAATAACAGCA      | C | T | Tyr | Tyr | SILENT-CODING | UNCLASSIFIED | Human Gene SWISSNEW-<br>ACC:P11245 ARYLAMINE N-<br>ACETYLTRANSFERASE,<br>POLYMORPHIC (EC 2.3.1.5)<br>(PNAT) (NAT-2) (ARYLAMINE<br>ACETYLASE) - Homo sapiens<br>(Human), 314 aa. | 5.40E-157 | <sup>8</sup><br>(8p23.1)         |
| 189 | cg43982075 | 499  | CTGCCATCTTTCAG<br>CCCTCTGAAAC[CT<br>JGTGTCCAGCACAG<br>AATCTTCCCTGG       | C | T | Thr | Thr | SILENT-CODING | UNCLASSIFIED | Human Gene SPTREMBL-<br>ACC:Q15729 THYROTROPH<br>EMBRYONIC FACTOR - HOMO<br>SAPIENS (HUMAN), 303 aa.  | 1.20E-154 | 22                               |
| 190 | cg43942977 | 350  | GCGCTTCCAGGT<br>CGGGACAATTCTG[G<br>T]CAGACTATTGIC<br>AAACTGGGAATA        | G | T | Arg | Arg | SILENT-CODING | UNCLASSIFIED | Human Gene Homologous to<br>SWISSNEW-ACC:Q12846<br>SYNTAXIN 4 - Homo sapiens<br>(Human), 297 aa.  | 9.60E-148 |                                  |
| 191 | cg43942977 | 701  | GGCAGCTGAAGAT<br>CACCAATGCTGG[G<br>C]ATGGTGTCTGAT<br>GAGGAGTGGAGC        | G | C | Gly | Gly | SILENT-CODING | UNCLASSIFIED | Human Gene Homologous to<br>SWISSNEW-ACC:Q12846<br>SYNTAXIN 4 - Homo sapiens<br>(Human), 297 aa.  | 9.60E-148 |                                  |

|     |            |     |   |   |   |     |               |              |   |           |    |
|-----|------------|-----|---|---|---|-----|---------------|--------------|---|-----------|----|
| 192 | cg43942977 | 773 | GCGAGGGTGTGT<br>GTC CAAT ATCCCTG/<br>TAAGGACACGCA<br>GGTGACTCGACAG<br>G | G | T | Leu | SILENT-CODING | UNCLASSIFIED | Human Gene Homologous to<br>SWISSPROT-ACC:Q12846<br>SYNTAXIN 4 - Homo sapiens<br>(Human), 297 aa.   | 9.60E-148 |    |
| 193 | cg43985220 | 753 | TGTACACTGCCAG<br>AAAAGGAAAGG/<br>TGIGCCTTTGTA<br>TGGTCAAAGACTA<br>C     | T | G | Gly | SILENT-CODING | UNCLASSIFIED | Human Gene Homologous to<br>SWISSPROT-ACC:P29218 MYO-<br>INOSITOL-1(OR 4)-<br>MONOPHOSPHATASE (EC<br>3.1.3.25) (IMP) (INOSITOL<br>MONOPHOSPHATASE)<br>(LITHIUM-SENSITIVE MYO-<br>INOSITOL<br>MONOPHOSPHATASE A1) -<br>Homo sapiens (Human), 277 aa. | 5.10E-145 | 8  |
| 194 | cg43985220 | 837 | TCTGGTGA<br>GTTGGGCTCTTC<br>TAGAACCCAGA<br>GACTGTGAGAAATG<br>G          | C | T | Ser | SILENT-CODING | UNCLASSIFIED | Human Gene Homologous to<br>SWISSPROT-ACC:P29218 MYO-<br>INOSITOL-1(OR 4)-<br>MONOPHOSPHATASE (EC<br>3.1.3.25) (IMP) (INOSITOL<br>MONOPHOSPHATASE)<br>(LITHIUM-SENSITIVE MYO-<br>INOSITOL<br>MONOPHOSPHATASE A1) -<br>Homo sapiens (Human), 277 aa. | 5.10E-145 | 8  |
| 195 | cg43946394 | 321 | TAGAGCGCACACA<br>GGCCTCCAGCTGIA<br>/GIGCCATGTCGGTC<br>TCATCATCCAAG      | A | G | Ala | SILENT-CODING | UNCLASSIFIED | Human Gene Homologous to<br>SWISSPROT-ACC:P29692<br>ELONGATION FACTOR 1-<br>DELTA (EF-1-DELTA) - Homo<br>sapiens (Human), 281 aa.   | 2.80E-144 | 19 |

|     |            |      |  |   |   |     |               |              |   |           |                 |
|-----|------------|------|--|---|---|-----|---------------|--------------|---|-----------|-----------------|
| 196 | cg43119818 | 1329 | CTGACAGCTACAG<br>GCTCTTCAAGTC/<br>TCAATTCACTGG<br>GGCAGTACAATG         | C | T | Phe | SILENT-CODING | UNCLASSIFIED | Human Gene Homologous to<br>SWISSPROT-ACC: P00915<br>CARBONIC ANHYDRASE I (EC<br>4.2.1.1) (CARBONATE<br>DEHYDRATASE I) - Homo<br>sapiens (Human), 260 aa. | 6.90E-141 | 8 (8q22)        |
| 197 | cg43118279 | 735  | AGAAGTTGAAGGG<br>GCTGGCTGCCACTV/<br>GJGGACCCGAATCA<br>AGTCGACACACTA    | T | G | Leu | SILENT-CODING | UNCLASSIFIED | Human Gene Homologous to<br>SWISSPROT-ACC: Q05195 MAD<br>PROTEIN (MAX DIMERIZER)-<br>Homo sapiens (Human), 221 aa.  | 1.20E-111 | 2 (2p13)        |
| 198 | cg43325007 | 866  | TGGGTCAAGGGAT<br>GTAGGCCCTCTCT/<br>CJACAGCCAGGCG<br>GCTCAGGGAAAC<br>A  | T | C | Val | SILENT-CODING | UNCLASSIFIED | Human Gene Homologous to<br>TREMBL NEW-ACC: AAD43195<br>PEROXISOMAL MEMBRANE<br>PROTEIN FMP 24 - HOMO<br>SAPIENS (HUMAN), 212 aa.                         | 4.80E-110 | 20              |
| 199 | cg39524111 | 402  | GCCAATAAGGAT<br>AGGGCACTACAGI<br>A/GTTCCGGTACA<br>GTGACACCCCTGGA<br>GC | A | G | Arg | SILENT-CODING | UNCLASSIFIED | Human Gene Similar to<br>TREMBL NEW-ACC: BAA13472<br>CD89 U08 - HOMO SAPIENS<br>(HUMAN), 191 aa.  | 2.10E-100 | 19<br>(19q13.4) |
| 200 | cg43280516 | 629  | ACGGGGAGGAGCT<br>GCAGATGAAACC/C<br>/T]GTGTAGGGTGC<br>TTCTGGGACCTGC     | C | T | Pro | SILENT-CODING | UNCLASSIFIED | Human Gene Similar to<br>TREMBL NEW-ACC: CAB43107<br>PRENYLATED RAB ACCEPTOR<br>1 (PRA1) - HOMO SAPIENS<br>(HUMAN), 185 aa.                               | 6.80E-95  | 19              |

|     |            |     |  |        |     |               |              |  |          |              |
|-----|------------|-----|--|--------|-----|---------------|--------------|--|----------|--------------|
| 201 | cg43963913 | 871 | AGAGGTGGGG<br>CGCCGAGGGCGAI<br>G/AICGGCCCCGAA<br>AGGGCTGGCTC<br>CT       | G<br>A | Arg | SILENT-CODING | UNCLASSIFIED | Human Gene Similar to SPTREMBL-ACC:O14803 BCL-X/BCL-2 BINDING PROTEIN - HOMO SAPIENS (HUMAN), 168 aa (fragment).                                 | 5.10E-90 | 11           |
| 202 | cg40262905 | 682 | TAGTGAAGGGCT<br>GAAATATATGCTTG<br>/C]GAGGTGGAAAT<br>TGGCAGAACTACC<br>T   | G<br>C | Leu | SILENT-CODING | UNCLASSIFIED | Human Gene Similar to TREMBLNEW-ACC:BAA34941 HUMAN CMAP - HOMO SAPIENS (HUMAN), 167 aa.  | 1.30E-89 |              |
| 203 | cg43918168 | 915 | CCTCATCAACAGC<br>ATCCGGACTGCCAT<br>/C]GGGGCTCGCC<br>GTGGGGCTGGGC<br>C    | T<br>C | Pro | SILENT-CODING | UNCLASSIFIED | Human Gene Similar to SWISSPROT-ACC:P09496 CLATHRINLIGHT CHAIN A (BRAIN AND LYMPHOCYTE LCA) - Homo sapiens (Human), 248 aa.                      | 3.80E-85 | 9<br>(12q23) |
| 204 | cg43259701 | 136 | CGACGAGGTGCTA<br>CGCGAGGGCGAGI<br>C/T]TGGAGAACGCG<br>CAGCGACAGCCTC<br>TT | C<br>T | Leu | SILENT-CODING | UNCLASSIFIED | Human Gene Similar to SPTREMBL-ACC:O00496 IPL (IPL) - HOMO SAPIENS (HUMAN), 152 aa.  | 1.30E-77 | 11           |
| 205 | cg1527767  | 162 | TTTTTCAGCTTA<br>CAATGGTACAGIA<br>GICAGGGAGCTGG<br>GGAAGGTCCTGTC<br>C     | A<br>G | Arg | SILENT-CODING | UNCLASSIFIED | Human Gene Similar to REMTREMBL-ACC:G36907 T-CELL RECEPTOR ALPHA-CHAIN HAP58 V(A)0.1-(A) <sup>T</sup> - HOMO SAPIENS (HUMAN), 135 aa (fragment). | 5.60E-68 |              |

|     |            |      |  |   |     |     |                  |                   |   |          |                 |
|-----|------------|------|--|---|-----|-----|------------------|-------------------|---|----------|-----------------|
| 206 | cg40968986 | 316  | AGAAGAGAGGCCCTG<br>TGACACTGGCAC[C<br>T]INGTGACTCAT<br>CGGCTGGCAGGCT      | C | T   | Thr | SILENT-CODING    | UNCLASSIFIED      | Human Gene Similar to<br>SWISSNEW-ACC:P06881<br>CALCITONIN GENE-RELATED<br>PEPTIDE 1 PRECURSOR (CGRP-<br>1) (ALPHA-TYPE CGRP) - Homo<br>sapiens (Human), 128 aa.        | 5.10E-58 | 11<br>(11p15.2) |
| 207 | cg42550133 | 300  | TCATCCTGAGTCT<br>AAGAACGCTCCT[T/<br>C]CTCAGTGA<br>GGCTCTTA<br>CTCT       | T | C   | Leu | SILENT-CODING    | UNCLASSIFIED      | Human Gene Similar to<br>REMTREMBL-ACC:D1002898 T-<br>CELL RECEPTOR BETA-CHAIN<br>V REGION - HOMO SAPIENS<br>(HUMAN), 112 aa (fragment).                                | 8.50E-56 | 7 (7q35)        |
| 208 | cg2526759  | 317  | CTCTGGTGTCCAC<br>GAGGGAGACACT/<br>CGTAACTCTCAAT<br>TGCAATTATGAAG         | T | C   | Thr | SILENT-CODING    | UNCLASSIFIED      | Human Gene Similar to<br>REMTREMBL-ACC:G33509 T<br>CELL RECEPTOR - HOMO<br>SAPIENS (HUMAN), 118 aa<br>(fragment).   | 1.60E-54 |                 |
| 209 | cg41664708 | 249  | AAGTCTGTGCTGA<br>TCCACAAGGCCAC[A<br>/G]TGGGTGAGAGA<br>CGTGGTCAGGAGC<br>A | A | G   | Thr | SILENT-CODING    | UNCLASSIFIED      | Human Gene Similar to<br>SWISSNEW-ACC:P47992<br>LYMPHOTACTIN PRECURSOR<br>(CYTOKINE SCM-1) (ATAC)<br>(LYMPHOTAXIN) (SCM-1-<br>ALPHA) - Homo sapiens (Human),<br>114 aa. | 2.00E-54 | 1               |
| 210 | cg43300673 | 1571 | AGGGAGGGGGGA<br>GGTAGCATGGG<br>G/gap]CACACGGCC<br>CTCACAGGGACTC<br>ACT   | G | gap |     | SILENT-NONCODING | ATPase_associated | Human Gene SPTREMBL-<br>ID:Q93050 VACUOLAR-TYPE<br>H(+)-ATPASE 115 KDA<br>SUBUNIT - HOMO SAPIENS<br>(HUMAN), 831 aa.  | 0        | 17              |

|     |            |      |   |     |   |  |                          |                       |   |           |    |
|-----|------------|------|---|-----|---|--|--------------------------|-----------------------|---|-----------|----|
| 211 | cg43284434 | 2570 | AGTTGAATCAGA<br>GAGGAATAAAAG<br>ap/A[GACATTATTA<br>ATTATTATTCTGCTC<br>C | gap | A |  | SILENT-<br>NONCOD<br>ING | ATPase_associat<br>ed | Human Gene Homologous to<br>SPTREMBL-ID:Q18788 C52E4.5<br>- CAENORHABDITIS<br>ELEGANS, 590 aa.  | 4.00E-121 | 6  |
| 212 | cg43132502 | 196  | TAAGCATGAGGTG<br>GCACCGAGGCAGG<br>[A/C]GGTGGCGATG<br>CCACCTGGGGTC<br>AC | A   | C |  | SILENT-<br>NONCOD<br>ING | ATPase_associat<br>ed | Human Gene Similar to<br>SPTREMBL-ID:Q15332 GAMMA<br>SUBUNIT OF SODIUM<br>POTASSIUM ATPASE LIKE -<br>HOMO SAPIENS (HUMAN), 126<br>aa. | 9.40E-58  | 11 |
| 213 | cg43931765 | 606  | GGTCCCCCTTGCTTT<br>ATCCCAGCTCGT<br>[GAGGGACGCCAGC<br>CTGGCATGGCTCT      | G   | T |  | SILENT-<br>NONCOD<br>ING | cadherin              | Human Gene SWISSPROT-<br>ID:P18084 INTEGRIN BETA-5<br>SUBUNIT PRECURSOR - HOMO<br>SAPIENS (HUMAN), 799 aa.                            | 0         | 3  |
| 214 | cg43931765 | 607  | GTCCCCTTGCTTA<br>TCCCAAGCTCGT<br>[AGGGACGCCAGC<br>GGCATGGCTCTG          | G   | T |  | SILENT-<br>NONCOD<br>ING | cadherin              | Human Gene SWISSPROT-<br>ID:P18084 INTEGRIN BETA-5<br>SUBUNIT PRECURSOR - HOMO<br>SAPIENS (HUMAN), 799 aa.                            | 0         | 3  |
| 215 | cg43931765 | 615  | CTTATCCCAAGCT<br>CGGAGGGACGCC<br>pG]AGCCTGGCATG<br>A                    | gap | G |  | SILENT-<br>NONCOD<br>ING | cadherin              | Human Gene SWISSPROT-<br>ID:P18084 INTEGRIN BETA-5<br>SUBUNIT PRECURSOR - HOMO<br>SAPIENS (HUMAN), 799 aa.                            | 0         | 3  |

|     |            |     |   |     |   |  |                           |          |  |   |   |
|-----|------------|-----|---|-----|---|--|---------------------------|----------|--|---|---|
| 216 | cg43931765 | 660 | TAGCAGCCAGGTG<br>ACATGGCCAGGC <sup>b</sup><br>ap/TACCTTCC <sup>c</sup><br>ACAGGCACTGTGG<br>GC   | gap | T |  | SILENT-<br>NONCOD-<br>ING | cadherin | Human Gene SWISSPROT-<br>ID:P18084 INTEGRIN BETA-5<br>SUBUNIT PRECURSOR - HOMO<br>SAPIENS (HUMAN), 799 aa. | 0 | 3 |
| 217 | cg43931765 | 665 | GCCAGGTGACATG<br>GCCAGGCCACCTT <sup>b</sup><br>ap/TJCCGTACAGG<br>CACTGTGGCTCC <sup>c</sup><br>G | gap | T |  | SILENT-<br>NONCOD-<br>ING | cadherin | Human Gene SWISSPROT-<br>ID:P18084 INTEGRIN BETA-5<br>SUBUNIT PRECURSOR - HOMO<br>SAPIENS (HUMAN), 799 aa. | 0 | 3 |
| 218 | cg43931765 | 668 | AGGTGACATGGCC<br>AGGCACCTTCCT <sup>b</sup><br>p/TJGTACAGGCACT<br>GTGGCTCC <sup>c</sup><br>C     | gap | T |  | SILENT-<br>NONCOD-<br>ING | cadherin | Human Gene SWISSPROT-<br>ID:P18084 INTEGRIN BETA-5<br>SUBUNIT PRECURSOR - HOMO<br>SAPIENS (HUMAN), 799 aa. | 0 | 3 |
| 219 | cg43931765 | 668 | AGGTGACATGGCC<br>AGGCACCTTCCT <sup>b</sup><br>p/TJGTACAGGCACT<br>GTGGCTCC <sup>c</sup><br>C     | gap | T |  | SILENT-<br>NONCOD-<br>ING | cadherin | Human Gene SWISSPROT-<br>ID:P18084 INTEGRIN BETA-5<br>SUBUNIT PRECURSOR - HOMO<br>SAPIENS (HUMAN), 799 aa. | 0 | 3 |
| 220 | cg43931765 | 668 | AGGTGACATGGCC<br>AGGCACCTTCCT <sup>b</sup><br>p/TJGTACAGGCACT<br>GTGGCTCC <sup>c</sup><br>C     | gap | T |  | SILENT-<br>NONCOD-<br>ING | cadherin | Human Gene SWISSPROT-<br>ID:P18084 INTEGRIN BETA-5<br>SUBUNIT PRECURSOR - HOMO<br>SAPIENS (HUMAN), 799 aa. | 0 | 3 |

|     |            |      |   |   |   |  |                           |          |  |           |                     |
|-----|------------|------|---|---|---|--|---------------------------|----------|--|-----------|---------------------|
| 221 | cg43952088 | 4769 | AATCCACAATTCGG<br>CATCAGGAAGCC[A<br> C]AAGTCCAGTG<br>GCCATTAGGGTCC<br>T | A | C |  | SILENT-<br>NONCOD-<br>ING | cadherin | Human Gene SPTREMBL-<br>ID:Q15065 OB-CADHERIN-1-<br>HOMO SAPIENS (HUMAN), 796<br>aa.   | 0         | 16                  |
| 222 | cg44010957 | 1406 | TCCCTATGAGCCTG<br>CAAAGGGAGACA[<br>T]TCAGGAATGAGT<br>TCCATGTCCGAGA      | G | T |  | SILENT-<br>NONCOD-<br>ING | cadherin | Human Gene SWISSPROT-<br>ID:P20701 LEUKOCYTE<br>ADHESION GLYCOPROTEIN<br>LFA-1 ALPHA CHAIN<br>PRECURSOR (LEUKOCYTE<br>FUNCTION ASSOCIATED<br>MOLECULE 1, ALPHA CHAIN)<br>(CD11A) (INTEGRIN ALPHA-L)<br>-HOMO SAPIENS (HUMAN),<br>1170 aa.  | 0         | 16<br>(16p11.2<br>) |
| 223 | cg43956560 | 1463 | CAGTGCATCTGGG<br>AAGATTCTACCT/<br>CTGACCAACAGITC<br>CTTCAGCTTCCAT       | T | C |  | SILENT-<br>NONCOD-<br>ING | cadherin | Human Gene SWISSPROT-<br>ID:P14151 L-SELECTIN<br>PRECURSOR (LYMPH NODE<br>HOMING RECEPTOR)<br>(LEUKOCYTE ADhesion<br>MOLECULE-1) (LAM-1)<br>(LEUKOCYTE SURFACE<br>ANTIGEN LEU-8) (IQ1) (GP90-<br>MEL) (LEUKOCYTE-<br>ENDOTHELIAL CELL<br>ADHESION MOLECULE 1)<br>(LECAM1) (CD62L) - HOMO<br>SAPIENS (HUMAN), 372 aa. | 1.00E-218 | 1 (1423)            |

|     |            |      |  |   |                          |                          |  |  |           |          |
|-----|------------|------|--|---|--------------------------|--------------------------|--|--|-----------|----------|
| 224 | cg43956560 | 1492 | CAACAGTTCCTICA<br>GCTTCCATTTC[G/A<br>ICCCCTCAATTATC<br>CCTCAAACCCCCA | A | SILENT-<br>NONCOD<br>ING | cadherin                 | Human Gene SWISSPROT-<br>ID:P14151 L-SELECTIN<br>PRECURSOR (LYMPH NODE<br>HOMING RECEPTOR)<br>(LEUKOCYTE ADHESION<br>MOLECULE-1) (LAM-1)<br>(LEUKOCYTE SURFACE<br>ANTIGEN LEU-8) (TQ1) (GP90-<br>MEL) (LEUKOCYTE-<br>ENDOTHELIAL CELL<br>ADHESION MOLECULE 1)<br>(LECAM1) (CD62L) - HOMO<br>SAPIENS (HUMAN), 372 aa. | 1.00E-218  | I (1q23)  |          |
| 225 | cg43956560 | 2242 | TGCTCTCTTTCCC<br>CTGGCCCCAGAAC/<br>[C]CTTTATCCACT<br>TACCTAGATTCTA   | C | A                        | SILENT-<br>NONCOD<br>ING | cadherin   | Human Gene SWISSPROT-<br>ID:P14151 L-SELECTIN<br>PRECURSOR (LYMPH NODE<br>HOMING RECEPTOR)<br>(LEUKOCYTE ADHESION<br>MOLECULE-1) (LAM-1)<br>(LEUKOCYTE SURFACE<br>ANTIGEN LEU-8) (TQ1) (GP90-<br>MEL) (LEUKOCYTE-<br>ENDOTHELIAL CELL<br>ADHESION MOLECULE 1)<br>(LECAM1) (CD62L) - HOMO<br>SAPIENS (HUMAN), 372 aa. | 1.00E-218 | I (1q23) |

|     |            |      |   |        |  |                          |            |  |           |               |
|-----|------------|------|---|--------|--|--------------------------|------------|--|-----------|---------------|
| 226 | cg43264626 | 428  | TGGCCACAGTGAA<br>AAAGGTCAATGGGT<br>[A]GGAGAGAAGCA<br>AAGTAGGAAGGAT<br>C | T<br>A |  | SILENT-<br>NONCOD<br>ING | cathepsin  | Human Gene SWISSPROT-<br>ID:P43235 CATHEPSIN K<br>PRECURSOR (EC 3.4.22.38)<br>(CATHEPSIN O) (CATHEPSIN<br>X) (CATHEPSIN O2) - HOMO<br>SAPIENS (HUMAN), 329 aa. | 4.10E-183 | 1             |
| 227 | cg43011543 | 1972 | ACCGCACCTTTC<br>ACCGGTGGGG[C/<br>GCCAGTGAAGTT<br>TAACAAACTGCTG          | C<br>G |  | SILENT-<br>NONCOD<br>ING | collagen   | Human Gene SWISSPROT-<br>ID:P27658 COLLAGEN ALPHA<br>1(VII) CHAIN PRECURSOR<br>(ENDOTHELIAL COLLAGEN)-<br>HOMO SAPIENS (HUMAN), 744<br>aa.                     | 0         |               |
| 228 | cg43011543 | 2096 | CATAACCACGTICA<br>CTGCAAGGGGGI<br>C/GIAACGTGGGG<br>TTGCTCTATCAAG<br>A   | C<br>G |  | SILENT-<br>NONCOD<br>ING | collagen   | Human Gene SWISSPROT-<br>ID:P27658 COLLAGEN ALPHA<br>1(VIII) CHAIN PRECURSOR<br>(ENDOTHELIAL COLLAGEN)-<br>HOMO SAPIENS (HUMAN), 744<br>aa.                    | 0         |               |
| 229 | cg43933757 | 2346 | GAAACCCAGTAGG<br>CTCCTGGAGGCCA<br>/CTGGTCAGCTGC<br>TTGGAATCAGCA         | A<br>C |  | SILENT-<br>NONCOD<br>ING | complement | Human Gene SWISSPROT-<br>ID:P10643 COMPLEMENT<br>COMPONENT C7 PRECURSOR-<br>HOMO SAPIENS (HUMAN), 843<br>aa.   | 0         | 5 (5p13)      |
| 230 | cg41553795 | 64   | TGGTGGTGTACC<br>CTTGGCCCTCCA[C/<br>GAGTCCTGCCACC<br>CTGCTGCCAC          | C<br>G |  | SILENT-<br>NONCOD<br>ING | complement | Human Gene Homologous to<br>SWISSPROT-ID:P07360<br>COMPLEMENT C8 GAMMA<br>CHAIN PRECURSOR - HOMO<br>SAPIENS (HUMAN), 202 aa.                                   | 1.40E-104 | 9<br>(9p34.3) |

|     |            |      |  |        |                          |     |   |                              |
|-----|------------|------|--|--------|--------------------------|-----|---|------------------------------|
| 231 | cg42542496 | 168  | AGCCCTTCCACC<br>CGGATAGATTCT<br>TCACCCCTGGCC<br>GCCCTTGCCCCA         | C<br>T | SILENT-<br>NONCOD<br>ING | csf | Human Gene SWISSPROT-<br>ID:P40225 THROMBOPOEITIN<br>PRECURSOR<br>(MEGAKARYOCYTE COLONY<br>STIMULATING FACTOR) (C-<br>MPL LIGAND) (ML)<br>(MEGAKARYOCYTE GROWTH<br>AND DEVELOPMENT FACTOR)<br>(MGDF) - HOMO SAPIENS<br>(HUMAN), 353 aa. | 1.20E-189<br>3<br>(3q26.3)   |
| 232 | cg42542496 | 179  | ACCCGGATAAGATT<br>CCTCACCCCTGG[C/<br>T]CCGCCTTGGCC<br>CACCCCTACTCTGC | C<br>T | SILENT-<br>NONCOD<br>ING | csf | Human Gene SWISSPROT-<br>ID:P40225 THROMBOPOEITIN<br>PRECURSOR<br>(MEGAKARYOCYTE COLONY<br>STIMULATING FACTOR) (C-<br>MPL LIGAND) (ML)<br>(MEGAKARYOCYTE GROWTH<br>AND DEVELOPMENT FACTOR)<br>(MGDF) - HOMO SAPIENS<br>(HUMAN), 353 aa. | 1.20E-189<br>3<br>(3q26.3)   |
| 233 | cg41533258 | 1356 | GTGCCCTGGACATT<br>GCCTTGCTGGAACT<br>GGGGGACTGGGA<br>TGTGGGAGGGAGC    | C<br>T | SILENT-<br>NONCOD<br>ING | csf | Human Gene Homologous to<br>SWISSPROT-ID:P0919<br>GRANULOCYTE COLONY-<br>STIMULATING FACTOR<br>PRECURSOR (G-CSF)<br>(PLURIPOTENTIN) - HOMO<br>SAPIENS (HUMAN), 207 aa.  | 1.50E-107<br>17<br>(17q11.2) |

|     |           |     |  |     |   |                           |               |   |           |    |
|-----|-----------|-----|--|-----|---|---------------------------|---------------|---|-----------|----|
| 234 | cg2753430 | 657 | ACGACTTTGAGCC<br>TCGGCGATCCTT <sup>ta</sup><br>P/GAGTCCAACGIC<br>CAGCTCGTCTCTG | gap | G | SILENT-<br>NONCOD-<br>ING | csf           | Human Gene Similar to<br>SWISSNEW-ID:P08700<br>INTERLEUKIN-3 PRECURSOR<br>(IL-3) (MULTIPOENTIAL<br>COLONY-STIMULATING<br>FACTOR) (HEMATOPOETIC<br>GROWTH FACTOR) (P-CELL<br>STIMULATING FACTOR)<br>(MAST-CELL GROWTH<br>FACTOR) (MCGF) - HOMO<br>SAPIENS (HUMAN), 152<br>aa.]pcds;SWISSPROT-ID:P08700<br>INTERLEUKIN-3 PRECURSOR<br>(IL-3) (MULTIPOENTIAL<br>COLONY-STIMULATING<br>FACTOR) (HEMATOPOETIC<br>GROWTH FACTOR) (P-CELL<br>STIMULATING FACTOR)<br>(MAST-CELL GROWTH<br>FACTOR) (MCGF) - HOMO<br>SAPIENS (HUMAN), 152 aa. | 1.10E-77  | 5  |
| 235 | cg4036323 | 225 | TGGGGCTTAAAAAG<br>GGCAACCCGGCG<br>/CIGGACCCCTCC<br>CCTAGTCGGGG                 | G   | C | SILENT-<br>NONCOD-<br>ING | dehydrogenase | Human Gene SWISSPROT.<br>ID:P00367 GLUTAMATE<br>DEHYDROGENASE 1<br>PRECURSOR (EC 1.4.1.3) (GDH)<br>- HOMO SAPIENS (HUMAN),<br>558 aa.   | 5.80E-303 | 10 |

|     |            |      |  |  |   |  |                           |               |   |           |          |
|-----|------------|------|--|--|---|--|---------------------------|---------------|---|-----------|----------|
| 236 | cg43918671 | 766  | GAGAGACCATTA<br>CITACATCAAGTTC/<br>TGGTTTATAGACA<br>TTGAAATCATATC        | C  | T |  | SILENT-<br>NONCOD-<br>ING | dehydrogenase | Human Gene SPTREMBL-<br>ID:Q14131<br>DIHYDROLIPOAMIDE<br>DEHYDROGENASE - HOMO<br>SAPIENS (HUMAN), 511 aa.   | 5.10E-272 | 7 (7431) |
| 237 | cg43057018 | 1995 | AGTTTCATATACT<br>TTTCTCTCAC[gap]<br>GTTTTGTCTATGTT<br>GAAAATTTCCTG       | gap  | G |  | SILENT-<br>NONCOD-<br>ING | dehydrogenase | Human Gene SWISSNEW-<br>ID:P08319 ALCOHOL<br>DEHYDROGENASE CLASS II PI<br>CHAIN (EC 1.1.1.1) - HOMO<br>SAPIENS (HUMAN), 391<br>aa. pcis:SWISSPROT-ID:P08319<br>ALCOHOL DEHYDROGENASE<br>CLASS II PI CHAIN (EC 1.1.1.1)<br>- HOMO SAPIENS (HUMAN), 391<br>aa.  | 1.30E-209 | 4 (4422) |
| 238 | cg44005808 | 3691 | ACAAGACAGAAC<br>TGAAGTGGATCC<br>[gap]CIAAAGGTGCTC<br>AGAGAGCGGGCCC<br>GC | ACAAGACAGAAC<br>TGAAGTGGATCC<br>[gap]CIAAAGGTGCTC<br>AGAGAGCGGGCCC<br>GC | C |  | SILENT-<br>NONCOD-<br>ING | dna_ma_bind   | Human Gene SWISSNEW-<br>ID:P19838 NUCLEAR FACTOR<br>NF-KAPPA-B P105 SUBUNIT<br>(DNA-BINDING FACTOR KBFI)<br>(EBP-1) [CONTAINS:<br>NUCLEAR FACTOR NF-KAPPA-<br>B P50 SUBUNIT] - HOMO<br>SAPIENS (HUMAN), 969<br>aa. pcis:SWISSPROT-ID:P19838<br>NUCLEAR FACTOR NF-KAPPA-<br>B P105 SUBUNIT (CONTAINS:<br>NUCLEAR FACTOR NF-<br>KAPPA-B P50 SUBUNIT) (DNA-<br>BINDING FACTOR KBFI) (EBP-<br>1) - HOMO SAPIENS (HUMAN),<br>969 aa. | 0         |          |

|     |            |      |  |     |   |                          |              |  |           |
|-----|------------|------|--|-----|---|--------------------------|--------------|--|-----------|
| 239 | cg44005808 | 630  | TCTTCCCTCTCCAG<br>CCGGCAGGCCCG<br>/GJC GCC GCT TAGG<br>AGGGAGAGCCAC<br>C | gap | G | SILENT-<br>NONCOD<br>ING | dna_rna_bind | Human Gene SWISSNEW-<br>ID:P19838 NUCLEAR FACTOR<br>NF-KAPPA-B P105 SUBUNIT<br>(DNA-BINDING FACTOR KBFI)<br>(EBP-1) [CONTAINS:<br>NUCLEAR FACTOR NF-KAPPA-<br>B P50 SUBUNIT] - HOMO<br>SAPIENS (HUMAN), 969<br>aa.[pc]:SWISSPROT-ID:P19838<br>NUCLEAR FACTOR NF-KAPPA-<br>B P105 SUBUNIT (CONTAINS:<br>NUCLEAR FACTOR NF-<br>KAPPA-B P50 SUBUNIT) (DNA-<br>BINDING FACTOR KBFI) (EBP-<br>1) - HOMO SAPIENS (HUMAN),<br>969 aa. | 0         |
| 240 | cg43956159 | 1244 | TGGCGAGTCAGG<br>GTCA CCC ACATA<br>Agt/ATCCATGCC<br>CGGGTGCTATGCC<br>GC   | gap | A | SILENT-<br>NONCOD<br>ING | dna_rna_bind | Human Gene SPTREMBL-<br>ID:Q99612 DNA-BINDING<br>PROTEIN CPBP - HOMO<br>SAPIENS (HUMAN), 290 aa<br>(fragment).   | 1.40E-159 |
| 241 | cg43956159 | 1248 | GAGTCCAAGGGTCA<br>CCCACATACCAT[ga<br>p/TGGCACCAACGGGT<br>GCTATGCCCTCT    | gap | T | SILENT-<br>NONCOD<br>ING | dna_rna_bind | Human Gene SPTREMBL-<br>ID:Q99612 DNA-BINDING<br>PROTEIN CPBP - HOMO<br>SAPIENS (HUMAN), 290 aa<br>(fragment).   | 1.40E-159 |

|     |            |      |   |     |   |  |                           |             |   |                           |    |
|-----|------------|------|---|-----|---|--|---------------------------|-------------|---|---------------------------|----|
| 242 | cg43956159 | 1268 | TACCATGCAACCAC<br>GGGTGCTATGCCIG<br>/A]CTTCTTACAGGA<br>CCTTTTAGCCCT         | G   | A |  | SILENT-<br>NONCOD-<br>ING | dna_ma_bind | Human Gene SPTREMBL-<br>ID:Q99612 DNA-BINDING<br>PROTEIN CPBP - HOMO<br>SAPIENS (HUMAN), 290 aa<br>(fragment).  | 1.40E-159                 | 10 |
| 243 | cg43956159 | 1342 | CCTGGAGGCAACT<br>GGGTAGGGTGCA[<br>GIC]AACGGCATGC<br>TTTGGCTGGAAACA<br>CG    | G   | C |  | SILENT-<br>NONCOD-<br>ING | dna_ma_bind | Human Gene SPTREMBL-<br>ID:Q99612 DNA-BINDING<br>PROTEIN CPBP - HOMO<br>SAPIENS (HUMAN), 290 aa<br>(fragment).  | 1.40E-159                 | 10 |
| 244 | cg43956159 | 1364 | CAGAACGGCATGC<br>TTTGGCTGGAAAC[ga<br>p/C]ACGGCATCCCTC<br>CTTCCACGGCCGG<br>C | gap | C |  | SILENT-<br>NONCOD-<br>ING | dna_ma_bind | Human Gene SPTREMBL-<br>ID:Q99612 DNA-BINDING<br>PROTEIN CPBP - HOMO<br>SAPIENS (HUMAN), 290 aa<br>(fragment).  | 1.40E-159                 | 10 |
| 245 | cg43971258 | 471  | CAGAGCTAGCTCT<br>GGCTCTTCAAGGC/C/<br>TACAAGTTCACAG<br>TCCTTCGCTCCTG         | C   | T |  | SILENT-<br>NONCOD-<br>ING | dna_ma_bind | Human Gene Similar to<br>SWISSNEW-ID:Q02535 DNA-<br>BINDING PROTEIN INHIBITOR<br>ID-3 (ID-LIKE PROTEIN<br>INHIBITOR HLH 1R21) (HELIX-<br>LOOP-HELIX PROTEIN HEIR-1)<br>- HOMO SAPIENS (HUMAN),<br>119 aa. pcis:SWISSPROT-<br>ID:Q02535 DNA-BINDING<br>PROTEIN INHIBITOR ID-3 (ID-<br>LIKE PROTEIN INHIBITOR<br>HLH 1R21) (HELIX-LOOP-<br>HELIX PROTEIN HEIR-1)-<br>HOMO SAPIENS (HUMAN), 119<br>aa. | 1.30E-60<br>(1p36.13<br>) |    |

|     |            |      |   |          |                          |                        |   |                                |
|-----|------------|------|---|----------|--------------------------|------------------------|---|--------------------------------|
| 246 | cg43971258 | 508  | GTCCTTCGCTCCTG<br>AGCACCAGGT<br>CAAGTCTCAGGAA<br>GGGATTGGTGA      | T<br>C   | SILENT-<br>NONCOD<br>ING | dna_mra_bind_in<br>rib | Human Gene Similar to<br>SWISSNEW-ID:Q02535 DNA-<br>BINDING PROTEIN INHIBITOR<br>ID-3 (ID-LIKE PROTEIN<br>INHIBITOR HLH 1R21) (HELIX-<br>LOOP-HELIX PROTEIN HEIR-1)<br>- HOMO SAPIENS (HUMAN),<br>119 aa.[pcis:SWISSPROT-<br>ID:Q02535 DNA-BINDING<br>PROTEIN INHIBITOR ID-3 (ID-<br>LIKE PROTEIN INHIBITOR<br>HLH 1R21) (HELIX-LOOP-<br>HELIX PROTEIN HEIR-1)-<br>HOMO SAPIENS (HUMAN), 119<br>aa. | 1.30E-60<br>1<br>(1p36.13<br>) |
| 247 | cg43982507 | 3373 | GATACCTTGCGTG<br>GATCAAGCTCTG<br>/CTGTACTTGACCG<br>TTTTTATAATTACT | gap<br>C | SILENT-<br>NONCOD<br>ING | ephl                   | Human Gene SWISSPROT-<br>ID:P98155 VERY LOW-<br>DENSITY LIPOPROTEIN<br>RECEPTOR PRECURSOR<br>(VLDL RECEPTOR) - HOMO<br>SAPIENS (HUMAN), 873 aa.   | 0<br>9 (9p24)                  |
| 248 | cg43982507 | 3739 | CAAAAAAATTAT<br>AAACTAATTGTTG<br>^/GTTACGTATGAA<br>GATACTTGTGACCT | gap<br>G | SILENT-<br>NONCOD<br>ING | ephl                   | Human Gene SWISSPROT-<br>ID:P98155 VERY LOW-<br>DENSITY LIPOPROTEIN<br>RECEPTOR PRECURSOR<br>(VLDL RECEPTOR) - HOMO<br>SAPIENS (HUMAN), 873 aa.   | 0<br>9 (9p24)                  |
| 249 | cg43982507 | 514  | CCTCCCTTCCCC<br>TTTCCCCCTCCCA/C<br>GCCGCCACCTCT<br>TCCTCCCTTCGG   | A<br>C   | SILENT-<br>NONCOD<br>ING | ephl                   | Human Gene SWISSPROT-<br>ID:P98155 VERY LOW-<br>DENSITY LIPOPROTEIN<br>RECEPTOR PRECURSOR<br>(VLDL RECEPTOR) - HOMO<br>SAPIENS (HUMAN), 873 aa.   | 0<br>9 (9p24)                  |

|     |            |      |   |     |   |  |                           |          |  |           |               |
|-----|------------|------|---|-----|---|--|---------------------------|----------|--|-----------|---------------|
| 250 | cg41554010 | 1371 | CTGCCCTGCCACCT<br>GTCGTGTCGTgap/<br>TICCAAGAAGITC<br>TGGTATGAACTTG      | gap | T |  | SILENT-<br>NONCOD-<br>ING | eph      | Human Gene SWISSPROT-<br>ID:P06727 APOLIPOPROTEIN A-<br>IV PRECURSOR (APO-AIV) -<br>HOMO SAPIENS (HUMAN), 396<br>aa.[pcis:SWISSPROT;ID:P06727]<br>APOLIPOPROTEIN A-IV<br>PRECURSOR (APO-AIV) -<br>HOMO SAPIENS (HUMAN), 396<br>aa. | 1.80E-203 | 11<br>(11q23) |
| 251 | cg41554010 | 1371 | CTGCCCTGCCACCT<br>GTCGTGTCGTgap/<br>TICCAAGAAGITC<br>TGGTATGAACTTG      | gap | T |  | SILENT-<br>NONCOD-<br>ING | eph      | Human Gene SWISSPROT-<br>ID:P06727 APOLIPOPROTEIN A-<br>IV PRECURSOR (APO-AIV) -<br>HOMO SAPIENS (HUMAN), 396<br>aa.[pcis:SWISSPROT;ID:P06727]<br>APOLIPOPROTEIN A-IV<br>PRECURSOR (APO-AIV) -<br>HOMO SAPIENS (HUMAN), 396<br>aa. | 1.80E-203 | 11<br>(11q23) |
| 252 | cg43984905 | 2376 | TCCCCTCCAGGACT<br>AGGCTGGAGGAAT<br>G/CJCCAG/TGGGT<br>CCCCCTGAGTGG<br>GC | G   | C |  | SILENT-<br>NONCOD-<br>ING | esterase | Human Gene SWISSPROT-<br>ID:P51178 1-<br>PHOSPHATIDYLINOSITOL-4,5-<br>BISPHOSPHATE<br>PHOSPHODIESTERASE DELTA<br>1 (EC 3.14.11) (PLC-DELTA-1)<br>(PHOSPHOLIPASE C-DELTA-1)<br>(PLC-III) - HOMO SAPIENS<br>(HUMAN), 756 aa.         | 0         | 3             |

|     |            |      |  |   |   |                          |              |  |           |               |
|-----|------------|------|--|---|---|--------------------------|--------------|--|-----------|---------------|
| 253 | cg43984905 | 2440 | CACATGGGGGAC<br>AGGGCTGGTGG<br>/C/C/GCTCCCCAGCC<br>TCTTGCTCAAGAGC  | G | C | SILENT-<br>NONCOD<br>ING | esterase     | Human Gene SWISSPROT-<br>ID:P51178 1-<br>PHOSPHATIDYLINOSITOL-4,5-<br>BISPHOSPHATE<br>PHOSPHODIESTERASE DELTA<br>1 (EC 3.1.4.11) (PLC-DELTA-1)<br>(PHOSPHOLIPASE C-DELTA-1)<br>(PLC-III) - HOMO SAPIENS<br>(HUMAN), 756 aa.  | 0         | 3             |
| 254 | cg43992911 | 382  | CTAAAGTCGGAGT<br>ATCTCTCTCCAAG/<br>A]ATTTCACGTCCT<br>GGCGGCCGTTCCA | G | A | SILENT-<br>NONCOD<br>ING | glycoprotein | Human Gene SWISSPROT-<br>ID:P08183 MULTIRUG<br>RESISTANCE PROTEIN 1 (P-<br>GLYCOPROTEIN 1) - HOMO<br>SAPIENS (HUMAN), 1280 aa.   | 0         | 7             |
| 255 | cg43932434 | 267  | TITCTAGAGGGGG<br>TCIGTGAAGAT[G/<br>ATGTAACTAGTAC<br>ACCCCAACCCCCA  | G | A | SILENT-<br>NONCOD<br>ING | glycoprotein | Human Gene SWISSPROT-<br>ID:P16070 CD44 ANTIGEN<br>PRECURSOR (PHAGOCYTIC<br>GLYCPROTEIN 1) (PGP-1)<br>(HUTCH-1) (EXTRACELLULAR<br>MATRIX RECEPTOR-III)<br>(ECMR-1) (GP90<br>LYMPHOYTE<br>HOMING/ADHESION<br>RECEPTOR) (HERMES<br>ANTIGEN) (HYALURONATE<br>RECEPTOR) (HEPARAN<br>SULFATE PROTEOGLYCAN)<br>(EPICAN) (CDW44) - HOMO<br>SAPIENS (HUMAN), 742 aa. | 1.80E-195 | 11<br>(11per) |

|     |            |     |  |   |   |  |                           |              |   |           |                |
|-----|------------|-----|--|---|---|--|---------------------------|--------------|---|-----------|----------------|
| 256 | cg43932434 | 306 | CCCCAACCCCCAA<br>CCTCAAGTGGAAA[A<br>/GICAATGCCAGG<br>GATTAGGCTATGG<br>A  | A | G |  | SILENT-<br>NONCOD-<br>ING | glycoprotein | Human Gene SWISSPROT-<br>ID:P16070 CD44 ANTIGEN<br>PRECURSOR (PHAGOCYTIC<br>GLYCOPROTEIN I) (PGP-1)<br>(HUTCH-I) (EXTRACELLULAR<br>MATRIX RECEPTOR-III)<br>(ECMR-III) (GP90<br>LYMPHOYTE<br>HOMING/ADHESION<br>RECEPTOR) (HERMES<br>ANTIGEN) (HYALURONATE<br>RECEPTOR) (HEPARAN<br>SULFATE PROTEOGLYCAN)<br>(EPICAN) (CDW44) - HOMO<br>SAPIENS (HUMAN), 742 aa. | 1.80E-195 | II<br>(11pter) |
| 257 | cg43318219 | 366 | GCGCAGGTCAAGAG<br>GGGGCCCGCAGC]<br>A/G]GGCCTCCGCG<br>AGGTCCCCACGCC<br>GG | A | G |  | SILENT-<br>NONCOD-<br>ING | glycoprotein | Human Gene SWISSNEW-<br>ID:P15813 T-CELL SURFACE<br>GLYCOPROTEIN CD1D<br>PRECURSOR (CD1D ANTIGEN)<br>(R3G1) - HOMO SAPIENS<br>(HUMAN), 335<br>aa. Ipcis:SWISSPROT-ID:P15813<br>T-CELL SURFACE<br>GLYCOPROTEIN CD1D<br>PRECURSOR (CD1D ANTIGEN)<br>(R3G1) - HOMO SAPIENS<br>(HUMAN), 335 aa.   | 3.10E-185 | 1 (1q21)       |

|     |            |      |   |          |  |                          |              |   |          |          |
|-----|------------|------|---|----------|--|--------------------------|--------------|---|----------|----------|
| 258 | cg43967861 | 1954 | CTCTATACTGTACA<br>CTCACCCATAAT[T/g<br>ap]TCAAAACAAITA<br>CACCATGGTATAA<br>A | T<br>gap |  | SILENT-<br>NONCOD<br>ING | glycoprotein | Human Gene Similar to<br>SWISSPROT-ID:Q08878<br>FIBULIN-1, ISOFORM C<br>PRECURSOR (BASEMENT-<br>MEMBRANE PROTEIN 90) (BM-<br>90) - MUS MUSCULUS<br>(MOUSE), 683 aa. | 8.20E-67 | 2        |
| 259 | cg43967861 | 1955 | TCTATACTGTACAC<br>TCACCCATAAT[T/g<br>ap]CAAAACAAATTAC<br>ACCATGGTATAAA<br>G | T<br>gap |  | SILENT-<br>NONCOD<br>ING | glycoprotein | Human Gene Similar to<br>SWISSPROT-ID:Q08878<br>FIBULIN-1, ISOFORM C<br>PRECURSOR (BASEMENT-<br>MEMBRANE PROTEIN 90) (BM-<br>90) - MUS MUSCULUS<br>(MOUSE), 683 aa. | 8.20E-67 | 2        |
| 260 | cg43965366 | 1411 | GCGGAATAAGCCTG<br>GGTTGGAAAAG[IC<br>T]ATGTTTTGAAA<br>TATGTGGGATCTC          | C<br>T   |  | SILENT-<br>NONCOD<br>ING | glycoprotein | Human Gene Similar to<br>SWISSPROT-ID:P49222<br>ERYTHROCYTE MEMBRANE<br>PROTEIN BAND 4.2 (P4.2)<br>(PALLIDIN) - MUS MUSCULUS<br>(MOUSE), 690 aa.                    | 8.90E-61 | 6 (6p25) |
| 261 | cg43965366 | 385  | TACTGACCTAAAT<br>CACACCTAGACIA<br>T]TATCAGAGGGA<br>AATTCTGACCATA<br>A       | A<br>T   |  | SILENT-<br>NONCOD<br>ING | glycoprotein | Human Gene Similar to<br>SWISSPROT-ID:P49222<br>ERYTHROCYTE MEMBRANE<br>PROTEIN BAND 4.2 (P4.2)<br>(PALLIDIN) - MUS MUSCULUS<br>(MOUSE), 690 aa.                    | 8.90E-61 | 6 (6p25) |

|     |            |      |  |          |  |                          |              |   |           |    |
|-----|------------|------|--|----------|--|--------------------------|--------------|---|-----------|----|
| 262 | cg43322513 | 1255 | TGTCCCTTGAAGAA<br>CATGCACTTGGCIA<br>/GICGGAATGGCACAA<br>AGCAAAATGGTAGA     | A<br>G   |  | SILENT-<br>NONCOD<br>ING | glycoprotein | Human Gene Similar to<br>SWISSPROT-ID:P13983<br>EXTENSIN PRECURSOR (CELL-<br>WALL HYDROXYPROLINE-<br>RICH GLYCOPROTEIN) -<br>NICOTIANA TABACUM<br>(COMMON TOBACCO), 620 aa. | 3.30E-54  | 12 |
| 263 | cg41637704 | 1397 | CCCGCGCCCCAGT<br>AGGAGGCCCGCGIg<br>Ap/G]CCCAGCAGGT<br>GCGGCACGGCACGG<br>AG | gap<br>G |  | SILENT-<br>NONCOD<br>ING | homeobox     | Human Gene SWISSPROT-<br>ID:P50219 HOMEOBOX<br>PROTEIN HB9 - HOMO<br>SAPIENS (HUMAN), 401 aa.   | 1.20E-224 | 7  |
| 264 | cg41637704 | 1423 | CCAGCAGGTGCGG<br>CGCGCACGGAGCig<br>Ap/G]CGCCGGCGG<br>CGGCTTCTCCGG<br>AG    | gap<br>G |  | SILENT-<br>NONCOD<br>ING | homeobox     | Human Gene SWISSPROT-<br>ID:P50219 HOMEOBOX<br>PROTEIN HB9 - HOMO<br>SAPIENS (HUMAN), 401 aa.   | 1.20E-224 | 7  |
| 265 | cg41637704 | 1817 | TGAAACTTGAAAC<br>CGCCTCTGGAGC[<br>T]GCCATTCTGCAG<br>AGTATTGAAAAA           | C<br>T   |  | SILENT-<br>NONCOD<br>ING | homeobox     | Human Gene SWISSPROT-<br>ID:P50219 HOMEOBOX<br>PROTEIN HB9 - HOMO<br>SAPIENS (HUMAN), 401 aa.   | 1.20E-224 | 7  |
| 266 | cg43980506 | 939  | TCCAAGAAAGGGT<br>CATGGAAAGCTTA[T<br>C]TGGGAATAATC<br>CTCTCAATTAGAA<br>A    | T<br>C   |  | SILENT-<br>NONCOD<br>ING | homeobox     | Human Gene TREMBLNEW-<br>ID:G2896172 LIM HOMEOBOX<br>PROTEIN COFACTOR - HOMO<br>SAPIENS (HUMAN), 373 aa.  | 1.60E-206 |    |

|     |            |      |  |                    |                           |                |   |           |               |
|-----|------------|------|--|--------------------|---------------------------|----------------|---|-----------|---------------|
| 267 | cg43961305 | 100  | GGGGGGTTTTTTT<br>TTTTTCTCTG[G/T]<br>TTTTTTTTTTT<br>TTTTTTTTT             | G<br>T<br>A<br>C   | SILENT-<br>NONCOD-<br>ING | hydrolase      | Human Gene SWISSPROT-<br>ID:P37980 INORGANIC<br>PYROPHOSPHATASE (EC<br>3.6.1.1) (PYROPHOSPHATE<br>PHOSPHO-HYDROLASE)<br>(PPASE) - BOS TAURUS<br>(BOVINE), 289 aa.   | 1.30E-156 | 10            |
| 268 | cg43998672 | 503  | CTGGGGTTTTCG<br>GGGAGGAACCAA<br>G/gap]GGCTCACGG<br>AGCCTCCCTGTGCTG<br>CA | G<br>gap<br>G<br>A | SILENT-<br>NONCOD-<br>ING | hydroxysteroid | Human Gene SPTREMBL-<br>ID:Q13194 11-BETA-<br>HYDROXYSTEROID<br>DEHYDROGENASE TYPE 2 -<br>HOMO SAPIENS (HUMAN), 405<br>aa.  | 2.00E-220 | 16<br>(16q22) |
| 269 | cg43998672 | 505  | GGGGGITTCGGG<br>GAGGAACCAAGG<br>G/gap]CTCACGGAG<br>CCCTCTGCTGCA<br>GT    | G<br>gap<br>G<br>A | SILENT-<br>NONCOD-<br>ING | hydroxysteroid | Human Gene SPTREMBL-<br>ID:Q13194 11-BETA-<br>HYDROXYSTEROID<br>DEHYDROGENASE TYPE 2 -<br>HOMO SAPIENS (HUMAN), 405<br>aa.  | 2.00E-220 | 16<br>(16q22) |
| 270 | cg42908571 | 1031 | GAGTTAAATTATGT<br>AAGTCATATTGag/<br>TATATTAAAGA<br>AGTACCACTGAA          | T<br>A<br>C<br>G   | SILENT-<br>NONCOD-<br>ING | interleukin    | Human Gene Homologous to<br>SWISSPROT-ID:P05231<br>INTERLEUKIN-6 PRECURSOR<br>(IL-6) (B-CELL STIMULATORY<br>FACTOR 2) (BSF-2)<br>(INTERFERON BETA-2)<br>(HYBRIDOMA GROWTH<br>FACTOR) - HOMO SAPIENS<br>(HUMAN), 212 aa. | 3.40E-108 | 7 (7p21)      |

|     |            |      |  |     |   |  |                           |                      |  |           |          |
|-----|------------|------|--|-----|---|--|---------------------------|----------------------|--|-----------|----------|
| 271 | cgg2908571 | 1178 | CCTACCTAAATA<br>AATGGCTAACTTGA<br>TTATACATATTIT<br>TAAAGAAATATT<br>A | gap | T |  | SILENT-<br>NONCOD-<br>ING | interleukin          | Human Gene Homologous to<br>SWISSPROT-ID:P05231<br>INTERLEUKIN-6 PRECURSOR<br>(IL-6) (B-CELL STIMULATORY<br>FACTOR 2) (BSF-2)<br>(INTERFERON BET-A-2)<br>(HYBRIDOMA GROWTH<br>FACTOR) - HOMO SAPIENS<br>(HUMAN), 212 aa. | 3.40E-108 | 7 (7p21) |
| 272 | cgg2164914 | 1617 | CAGCCCCATTTG<br>GGTCACAGGAAGT<br>CJAGAGGAGGCCA<br>CGTCTCTACTAGTT     | T   | C |  | SILENT-<br>NONCOD-<br>ING | interleukin receptor | Human Gene SWISSPROT-<br>ID:P25025 HIGH AFFINITY<br>INTERLEUKIN-8 RECEPTOR B<br>(IL-8R B) (CXCR-2) (GROMGSA<br>RECEPTOR) (IL-8 RECEPTOR<br>TYPE 2) - HOMO SAPIENS<br>(HUMAN), 360 aa.                                    | 9.60E-191 | 2 (2q35) |
| 273 | cgg3958501 | 1133 | CCCAACCTGGTTT<br>GGCAGACATCA[A]<br>GAAATGATGGAGTA<br>CATTTTGAGATA    | A   | G |  | SILENT-<br>NONCOD-<br>ING | isomerase            | Human Gene SWISSPROT-<br>ID:P46926 PUTATIVE<br>GLUCOSAMINE-6-PHOSPHATE<br>ISOMERASE (EC 5.3.1.10)<br>(GLUCOSAMINE-6-<br>PHOSPHATE DEAMINASE)<br>(OSCILLIN) (KIAA0060)-<br>HOMO SAPIENS (HUMAN), 289<br>aa.               | 1.60E-156 | 5        |

|     |            |      |  |   |   |  |                           |           |   |           |          |
|-----|------------|------|--|---|---|--|---------------------------|-----------|---|-----------|----------|
| 274 | cg43958501 | 805  | CACCCAGGTCT<br>CCTAGTTAGA/G/<br>A/AAAAGCTGTGA<br>AAAGTGGAGAAAGG<br>A | G | A |  | SILENT-<br>NONCOD-<br>ING | isomerase | Human Gene SWISSPROT-<br>ID:P46926 PUTATIVE<br>GLUCOSAMINE-6-PHOSPHATE<br>ISOMERASE (EC 5.3.1.10)<br>(GLUCOSAMINE- 6-<br>PHOSPHATE DEAMINASE)<br>(OSCILLIN) (KIAA0060)-<br>HOMO SAPIENS (HUMAN), 289<br>aa.                                   | 1.60E-156 | 5        |
| 275 | cg43090990 | 2710 | TITATTCTATTCT<br>AICTGTGGATGT/G<br>IGTAAATGGCTGGG<br>GGGCCAGCCCCCTG  | T | G |  | SILENT-<br>NONCOD-<br>ING | kinase    | Human Gene SWISSPROT-<br>ID:Q04759 PROTEIN KINASE C <sub>z</sub><br>THETA TYPE (EC 2.7.1.-)<br>(NPKC-THETA) - HOMO<br>SAPIENS (HUMAN), 706 aa.  | 0         | 10       |
| 276 | cg42879455 | 2259 | AGCCTTTGGCTCC<br>CACTAAATACA/]<br>C/AAAGGCCCTCT<br>CTACATCTGGAA      | A | C |  | SILENT-<br>NONCOD-<br>ING | kinase    | Human Gene SWISSPROT-<br>ID:Q06187 TYROSINE-PROTEIN<br>KINASE BTK (EC 2.7.1.112)<br>(BRUTON'S TYROSINE<br>KINASE)<br>(AGAMMAGLOBULINAEMIA<br>TYROSINE KINASE) (ATK) (B<br>CELL PROGENITOR KINASE)<br>(BPK) - HOMO SAPIENS<br>(HUMAN), 659 aa. | 0         | X(q21.3) |

|     |            |      |   |     |   |                           |        |  |           |               |
|-----|------------|------|---|-----|---|---------------------------|--------|--|-----------|---------------|
| 277 | cg42879455 | 2283 | AAAAAGGCCCTC<br>TCTACATCTGGGA/<br>GATGCACCTCTTC<br>TTTGATTCCCTGG        | A   | G | SILENT-<br>NONCOD-<br>ING | kinase | Human Gene SWISSPROT-<br>ID:Q06187 TYROSINE-PROTEIN<br>KINASE BTK (EC 2.7.1.12)<br>(BRUTON'S TYROSINE<br>KINASE)<br>(AGAMMAGLOBULINAEMIA<br>TYROSINE KINASE) (ATK) (B<br>CELL PROGENITOR KINASE)<br>(BPK) - HOMO SAPIENS<br>(HUMAN), 659 aa. | 0         | X<br>(Xq21.3) |
| 278 | cg43971741 | 2151 | AGCAACTTGGCTG<br>AGCCCCAACATA[C<br>T]ACAGAGAAATC<br>ATCAAACCTGACTT<br>A | C   | T | SILENT-<br>NONCOD-<br>ING | kinase | Human Gene SPTREMBL-<br>ID:Q92749 TYPE I<br>PHOSPHATIDYLINOSITOL-4-<br>PHOSPHATE 5-KINASE BETA<br>(EC 2.7.1.68) (STM-7 PROTEIN)-<br>HOMO SAPIENS (HUMAN), 540<br>aa.   | 1.40E-290 | 9             |
| 279 | cg43971741 | 2200 | TAAGAGTTTCAA<br>GATGTCAAACTTC/<br>AAGGGCTGATCAGC<br>AGATGGGATGTGA       | C   | A | SILENT-<br>NONCOD-<br>ING | kinase | Human Gene SPTREMBL-<br>ID:Q92749 TYPE I<br>PHOSPHATIDYLINOSITOL-4-<br>PHOSPHATE 5-KINASE BETA<br>(EC 2.7.1.68) (STM-7 PROTEIN)-<br>HOMO SAPIENS (HUMAN), 540<br>aa.   | 1.40E-290 | 9             |
| 280 | cg43971741 | 2451 | TTTTAAAAATCCA<br>TCCACACACATgap<br>TGGTAAATTAAAG<br>TATAAAATCTTGT       | gap | T | SILENT-<br>NONCOD-<br>ING | kinase | Human Gene SPTREMBL-<br>ID:Q92749 TYPE I<br>PHOSPHATIDYLINOSITOL-4-<br>PHOSPHATE 5-KINASE BETA<br>(EC 2.7.1.68) (STM-7 PROTEIN)-<br>HOMO SAPIENS (HUMAN), 540<br>aa.   | 1.40E-290 | 9             |

|     |            |      |  |   |     |  |                           |        |  |           |    |
|-----|------------|------|--|---|-----|--|---------------------------|--------|--|-----------|----|
| 281 | cg43947749 | 1996 | AACGTCGATTTCGC<br>ACCGTCCAACCTTG<br>[gap]GCCCGCCCC<br>TCCTACAGCTGTA<br>AC    | G | gap |  | SILENT-<br>NONCOD-<br>ING | kinase | Human Gene SWISSPROT-<br>ID:P49840 GLYCOGEN<br>SYNTHASE KINASE-3 ALPHA<br>(EC 2.7.1.37) (GSK-3 ALPHA)-<br>HOMO SAPIENS (HUMAN), 483<br>aa.                   | 5.60E-267 | 19 |
| 282 | cg43947749 | 1997 | ACGTGCGATTTCGCA<br>CCGTCCAACCTTG[G<br>[gap]CCCCGGCCCTC<br>CTACAGCTGTAAC<br>T | G | gap |  | SILENT-<br>NONCOD-<br>ING | kinase | Human Gene SWISSPROT-<br>ID:P49840 GLYCOGEN<br>SYNTHASE KINASE-3 ALPHA<br>(EC 2.7.1.37) (GSK-3 ALPHA)-<br>HOMO SAPIENS (HUMAN), 483<br>aa.                   | 5.60E-267 | 19 |
| 283 | cg44131752 | 1535 | CACTTAATACCAAG<br>AGACCCCCCCCC[G<br>[gap]CTTCCCCCTCCCC<br>CTTCCCCCCCCCT      | C | gap |  | SILENT-<br>NONCOD-<br>ING | kinase | Human Gene SPTREMBL-<br>ID:Q15599 TYROSINE KINASE<br>ACTIVATOR PROTEIN 1 (TKA-<br>1) - HOMO SAPIENS (HUMAN),<br>450 aa.                                      | 7.80E-173 | 16 |
| 284 | cg43917718 | 306  | AGACGTGCTGCC<br>ACAGGTCTCAGGA<br>[G]TAACAGATGCC<br>CTGTCCACTGAGA<br>G        | A | G   |  | SILENT-<br>NONCOD-<br>ING | kinase | Human Gene Similar to<br>SPTREMBL-ID:Q15599<br>TYROSINE KINASE<br>ACTIVATOR PROTEIN 1 (TKA-<br>1) - HOMO SAPIENS (HUMAN),<br>450 aa.                         | 1.40E-79  | 17 |
| 285 | cg43928048 | 1876 | TTTGATGGAAAGG<br>TTGTCACACTTG[G<br>A]GAATTATCACAC<br>ACTGATCAGGAA            | G | A   |  | SILENT-<br>NONCOD-<br>ING | kinase | Human Gene Similar to<br>SWISSPROT-ID:P20505 30 KD<br>PROTEIN KINASE HOMOLOG<br>(EC 2.7.1.-) (PROTEIN B) -<br>VACCinia VIRUS (STRAIN<br>COPENHAGEN), 300 aa. | 5.30E-55  |    |

|     |            |      |  |   |   |  |                           |                 |   |          |                     |
|-----|------------|------|--|---|---|--|---------------------------|-----------------|---|----------|---------------------|
| 286 | cg42714751 | 208  | CCCTCCGGATTGCG<br>GCGCGCTGCGGC<br>/MCCGCCGAGT<br>GAGGGTTTCGTG<br>G | C | M |  | SILENT-<br>NONCOD-<br>ING | Kinaseinhibitor | Human Gene Similar to<br>SWISSPROT-ID:P42771<br>CYCLIN-DEPENDENT KINASE<br>4 INHIBITOR A (CDK4) (P16-<br>INK4) (P16-INK4A) (MULTIPLE<br>TUMOR SUPPRESSOR 1)<br>(MTS1) - HOMO SAPIENS<br>(HUMAN), 156 aa.  | 2.60E-53 | 9 (9p21)            |
| 287 | cg43322545 | 2943 | TCCAAGCTAACCA<br>CTGCCACTGGGA<br>/GAAACTCACCT<br>CCCACTTCCCAC      | A | G |  | SILENT-<br>NONCOD-<br>ING | Kinaserceptor   | Human Gene SWISSNEW-<br>ID:P30530 TYROSINE-PROTEIN<br>KINASE RECEPTOR UFO<br>PRECURSOR (EC 2.7.1.112)<br>(AXL ONCOGENE) - HOMO<br>SAPIENS (HUMAN), 887<br>aa.[pc]:SWISSPROT-ID:P30530<br>TYROSINE-PROTEIN KINASE<br>RECEPTOR UFO PRECURSOR<br>(EC 2.7.1.112) (AXL<br>ONCOGENE) - HOMO SAPIENS<br>(HUMAN), 887 aa. | 0        | 19<br>(19q13.1<br>) |

|     |            |      |  |   |   |                           |                |  |   |                     |
|-----|------------|------|--|---|---|---------------------------|----------------|--|---|---------------------|
| 288 | cg43322545 | 3037 | CCACCTCCATCCCA<br>GACAGGTCCCTC/<br>GCCCTTCCTCTGTG<br>CAGTAGCATCACC       | C | G | SILENT-<br>NONCOD-<br>ING | kinasereceptor | Human Gene SWISSNEW-<br>ID:P30530 TYROSINE-PROTEIN<br>KINASE RECEPTOR UFO<br>PRECURSOR (EC 2.7.1.112)<br>(AXL ONCOGENE) - HOMO<br>SAPIENS (HUMAN), 887<br>aa.lpcis:SWISSPROT-ID:P30530<br>TYROSINE-PROTEIN KINASE<br>RECEPTOR UFO PRECURSOR<br>(EC 2.7.1.112) (AXL<br>ONCOGENE) - HOMO SAPIENS<br>(HUMAN), 887 aa. | 0 | 19<br>(19q13.1<br>) |
| 289 | cg43322545 | 3038 | CACCTCCATCCCA<br>GACAGGTCCCTC/<br>GCCCTTCCTCTGTG<br>AGTAGCATCACCT        | C | G | SILENT-<br>NONCOD-<br>ING | kinasereceptor | Human Gene SWISSNEW-<br>ID:P30530 TYROSINE-PROTEIN<br>KINASE RECEPTOR UFO<br>PRECURSOR (EC 2.7.1.112)<br>(AXL ONCOGENE) - HOMO<br>SAPIENS (HUMAN), 887<br>aa.lpcis:SWISSPROT-ID:P30530<br>TYROSINE-PROTEIN KINASE<br>RECEPTOR UFO PRECURSOR<br>(EC 2.7.1.112) (AXL<br>ONCOGENE) - HOMO SAPIENS<br>(HUMAN), 887 aa. | 0 | 19<br>(19q13.1<br>) |
| 290 | cg43980494 | 1040 | GTCTGATAGAAGA<br>GGAGCAGGAGAA/<br>A/GJCAAATCGTTA<br>AAACCTAGCGAAAT<br>TC | A | G | SILENT-<br>NONCOD-<br>ING | kinesin        | Human Gene SPTRMBL-<br>ID:Q14807 KID (KINESIN-LIKE<br>DNA BINDING PROTEIN) -<br>HOMO SAPIENS (HUMAN), 665<br>aa.   | 0 | 16                  |

|     |            |      |   |   |   |  |                           |              |   |           |               |
|-----|------------|------|---|---|---|--|---------------------------|--------------|---|-----------|---------------|
| 291 | cg43925424 | 374  | TCAAGGAGCAAGGC<br>GAATGTATGACAIA<br>C[CATGTCACAAAT<br>GGTGTACATAAAG | A | C |  | SILENT-<br>NONCOD-<br>ING | kinesin      | Human Gene SWISSPROT-<br>ID:Q07866 KINESIN LIGHT<br>CHAIN (KLC) - HOMO SAPIENS<br>(HUMAN), 569 aa.  | 1.90E-304 | 14            |
| 292 | cg42479188 | 305  | TCTGAAGAGGCT<br>GACGATTACT[A/<br>GTTCTCATTTTTC<br>CTTCTCCAGAA       | A | G |  | SILENT-<br>NONCOD-<br>ING | MHC          | Human Gene Homologous to<br>SWISSPROT-ID:P13765 HLA<br>CLASS II<br>HISTOCOMPATIBILITY<br>ANTIGEN, D0 BETA CHAIN<br>PRECURSOR - HOMO SAPIENS<br>(HUMAN), 273 aa.                   | 3.40E-147 | 6<br>(6p21.3) |
| 293 | cg42686658 | 1167 | CTAGCTTCCCTCC<br>CATTCAAACACAIA/<br>CIACACACATTCTT<br>GCTCTACCAAAAG | A | C |  | SILENT-<br>NONCOD-<br>ING | MHC          | Human Gene Homologous to<br>SWISSPROT-ID:P06340 HLA<br>CLASS II<br>HISTOCOMPATIBILITY<br>ANTIGEN, D2 ALPHA CHAIN<br>PRECURSOR (MHCDN-ALPHA)<br>- HOMO SAPIENS (HUMAN),<br>250 aa. | 3.70E-134 | 6<br>(6p21.3) |
| 294 | cg38337333 | 1122 | TGTCCTAACCCA<br>GCCTGCCAGCTCT/<br>CIAATGTACCAAGCA<br>GCTGGAATCTGAA  | T | C |  | SILENT-<br>NONCOD-<br>ING | MHC          | Human Gene Homologous to<br>SPTREMBL-ID:Q95368 HLA<br>CLASS I INHIBITORY NK<br>RECEPTOR - HOMO SAPIENS<br>(HUMAN), 455 aa.  | 1.80E-113 | 19            |
| 295 | cg27803682 | 2506 | TGGGTGGGTAT<br>TCCATCCATCTGT/<br>GIAAGCACATTGA<br>GCCTCCAGGCTTC     | T | G |  | SILENT-<br>NONCOD-<br>ING | misc_channel | Human Gene Similar to<br>SPTREMBL-ID:P91197 SIMILAR<br>TO LIGAND-GATED IONIC<br>CHANNEL PROTEIN -<br>CAENORHABDITIS ELEGANS,<br>461 aa.   | 3.50E-81  |               |

|     |            |      |   |     |   |  |                          |              |   |          |
|-----|------------|------|---|-----|---|--|--------------------------|--------------|---|----------|
| 296 | cg21413267 | 1440 | CGAGGGGACCCA<br>GAGCCCTGACCCIT<br>/GICCTCACCGTCC<br>TCTGCTCCCC        | T   | G |  | SILENT-<br>NONCOD<br>ING | misc_channel | Human Gene Similar to<br>SPTREMBL-ID:P91197 SIMILAR<br>TO LIGAND-GATED IONIC<br>CHANNEL PROTEIN -<br>CAENORHABDITIS ELEGANS,<br>461 aa. | 7.90E-79 |
| 297 | cg21413267 | 1860 | AGGAGCCCTCTTC<br>GGTGTCCCCGAGT<br>[C]GCCACGGTCAA<br>GACCCGAGCACC<br>A | T   | C |  | SILENT-<br>NONCOD<br>ING | misc_channel | Human Gene Similar to<br>SPTREMBL-ID:P91197 SIMILAR<br>TO LIGAND-GATED IONIC<br>CHANNEL PROTEIN -<br>CAENORHABDITIS ELEGANS,<br>461 aa. | 7.90E-79 |
| 298 | cg21413267 | 1890 | CGGTCAAGACCCG<br>CAGCACCAAAGC<br>A/GICCGCCCCGC<br>ACCTGCCCCGTG<br>C   | A   | G |  | SILENT-<br>NONCOD<br>ING | misc_channel | Human Gene Similar to<br>SPTREMBL-ID:P91197 SIMILAR<br>TO LIGAND-GATED IONIC<br>CHANNEL PROTEIN -<br>CAENORHABDITIS ELEGANS,<br>461 aa. | 7.90E-79 |
| 299 | cg2481172  | 1541 | GAGCCGTGGCT<br>GTGGCCTCGGGIA<br>/C]GGCGGTGGACG<br>GCGTGCCTCAIC<br>C   | A   | C |  | SILENT-<br>NONCOD<br>ING | misc_channel | Human Gene Similar to<br>SPTREMBL-ID:P91197 SIMILAR<br>TO LIGAND-GATED IONIC<br>CHANNEL PROTEIN -<br>CAENORHABDITIS ELEGANS,<br>461 aa. | 2.30E-71 |
| 300 | cg29518465 | 89   | GGGTGCACGGCCG<br>GCCCTGGGCAGG<br>gap                                  | gap | C |  | SILENT-<br>NONCOD<br>ING | oncogene     | Human Gene SWISSPROT-<br>ID:P15498 VAV PROTO-<br>ONCOGENE - HOMO SAPIENS<br>(HUMAN), 846 aa.  | 0        |

|     |            |      |   |     |   |  |                          |          |  |           |              |
|-----|------------|------|---|-----|---|--|--------------------------|----------|--|-----------|--------------|
| 301 | cg41972699 | 627  | ATGGGGCCGGTGT<br>CTGCCAGGAGG<br>ap/CIGCAGACCGG<br>CTCCAGGCCAGC<br>GC    | gap | C |  | SILENT-<br>NONCOD<br>ING | oncogene | Human Gene Similar to<br>SWISSPROT-ID:Q64010 PROTO-<br>ONCOGENE C-CRK (F38)<br>(ADAPTER MOLECULE CRK)-<br>MUS MUSCULUS (MOUSE), 304<br>aa.   | 2.40E-84  | 22<br>(22q1) |
| 302 | cg40333812 | 235  | AGCATTTGAGGAA<br>GCATAACTGACG[C<br>/T]GTGAACGGGGT<br>GTGGGGTACTTGC<br>C | C   | T |  | SILENT-<br>NONCOD<br>ING | oncogene | Human Gene Similar to<br>SWISSPROT-ID:P31695<br>NEUROGENIC LOCUS NOTCH1<br>HOMOLOG PROTEIN 4<br>PRECURSOR (TRANSFORMING<br>PROTEIN INT-3) - MUS<br>MUSCULUS (MOUSE), 1964 aa.  | 1.40E-62  |              |
| 303 | cg43280482 | 2295 | AGCATCTGGAGAC<br>GACCCCCGCAGCA<br>/CITTTCCCTCGGAC<br>CCCCCTGAAAGCC      | A   | C |  | SILENT-<br>NONCOD<br>ING | oncogene | Human Gene Similar to<br>TREMBLNEW-ID:G2952331<br>ARGAB1-INTERACTING<br>PROTEIN ARGBP2A - HOMO<br>SAPIENS (HUMAN), 666 aa.   | 3.90E-62  | 8            |
| 304 | cg4014837  | 22   | CACTGCTGTGCAG<br>GGCAGGGAA[T]GC<br>TCCAGGCAGACAG<br>CCCAGCAAAG          | A   | T |  | SILENT-<br>NONCOD<br>ING | oxidase  | Human Gene SWISSNEW-<br>ID:P08684 CYTOCHROME P450<br>3A4 (EC 1.14.14.1) (CYP3A4)<br>(NIFEDIPINE OXIDASE) (NF-25)<br>(P450-PCN1) - HOMO SAPIENS<br>(HUMAN), 502<br>aa. Ipcis:SWISSPROT-ID:P08684<br>CYTOCHROME P450 3A4 (EC<br>1.14.14.1) (NIFEDIPINE<br>OXIDASE) (NF-25) (P450-PCN1)<br>- HOMO SAPIENS (HUMAN),<br>502 aa. | 8.00E-257 |              |

|     |            |      |  |   |     |  |                           |             |   |           |                          |
|-----|------------|------|--|---|-----|--|---------------------------|-------------|---|-----------|--------------------------|
| 305 | cgi1626506 | 3178 | CAGCACAGCGAGC<br>GCTCTCAATCTGIA/<br>gap]CCCTTTTCCCT<br>TTCTCAGCCAAT      | A | gap |  | SILENT-<br>NONCOD-<br>ING | peroxidase  | Human Gene SWISSPROT-<br>ID:P07202 THYROID<br>PEROXIDASE PRECURSOR (EC<br>1.11.1.8) (TPO) - HOMO<br>SAPIENS (HUMAN), 933 aa.  | 0         | <sup>3</sup><br>(3426.3) |
| 306 | cgi3918944 | 2958 | TCTGTAGAGCTCTG<br>AAAAGGTGACTT/<br>GJATATAGGGTCT<br>TGTATGTTTAC          | T | G   |  | SILENT-<br>NONCOD-<br>ING | phosphatase | Human Gene SPTREMBL-<br>ID:Q15172 PROTEIN<br>PHOSPHATASE 2A/B56-ALPHA<br>- HOMO SAPIENS (HUMAN),<br>486 aa.   | 4.60E-246 | 1                        |
| 307 | cgi3988365 | 1537 | GACAGACGAGACA<br>GTGAGGTATGTGIA/<br>G]GGCTGGCTCCGG<br>AATGGTCCGGAGG<br>C | A | G   |  | SILENT-<br>NONCOD-<br>ING | phosphatase | Human Gene SWISSPROT-<br>ID:Q14642 TYPE I INOSITOL-<br>1,4,5-TRIPHOSPHATE 5-<br>PHOSPHATASE (EC 3.1.3.56)<br>(SPTASE) - HOMO SAPIENS<br>(HUMAN), 412<br>aa.[pcis:SPTREMBL-ID:Q14642<br>INOSITOL 1,4,5-TRIPHOPHATE<br>5-PHOSPHATASE - HOMO<br>SAPIENS (HUMAN), 412 aa. | 2.60E-227 | 10                       |
| 308 | cgi3969460 | 581  | TAACATATGAAAGA<br>CAAGACTGGTC<br>G]TCACGTTGGCGT<br>CTCTAGTTGATT          | C | G   |  | SILENT-<br>NONCOD-<br>ING | phosphatase | Human Gene SWISSPROT-<br>ID:P36876 PROTEIN<br>PHOSPHATASE PP2A, 55 KD<br>REGULATORY SUBUNIT,<br>ALPHA ISOFORM (PROTEIN<br>PHOSPHATASE PP2A B<br>SUBUNIT ALPHA ISOFORM)<br>(ALPHA-PR55) - RATTUS<br>NORVEGICUS (RAT), 447 aa.  | 1.90E-202 |                          |

|     |            |      |  |        |  |                          |                       |  |                       |
|-----|------------|------|--|--------|--|--------------------------|-----------------------|--|-----------------------|
| 309 | cg43933809 | 362  | AATTAAACCTCTA<br>GGTGTATACTTAT/<br>CIACTGAACTAGT<br>TATTTCCTATTA   | T<br>C |  | SILENT-<br>NONCOD<br>ING | phosphatase           | Human Gene SWISSPROT-<br>ID:P37140 SERINE/THREONINE<br>PROTEIN PHOSPHATASE PP1-<br>BETA CATALYTIC SUBUNIT<br>(EC 3.1.3.16) (PP-1B) - HOMO<br>SAPIENS (HUMAN), RATTUS<br>NORVEGICUS (RAT), MUS<br>MUSCULUS (MOUSE),, 327 aa.  | 1.60E-181<br>2 (2p23) |
| 310 | cg43931444 | 215  | TGCTCGGCCGTG<br>CCACTAAGGTCAIC<br>TTJCCCCGCTCCGQA<br>GAGCCCAGAGCCG | C<br>T |  | SILENT-<br>NONCOD<br>ING | phosphataseinhi<br>b  | Human Gene Similar to<br>SWISSPROT-ID:P39687 POTENT<br>HEAT-STABLE PROTEIN<br>PHOSPHATASE 2A INHIBITOR<br>IIPIP2A (HLA-DR ASSOCIATED<br>PROTEIN 1) (PHAP) (ACIDIC<br>NUCLEAR PHOSPHOPROTEIN<br>PP32) (CEREBELLAR LEUCINE<br>RICH ACIDIC NUCLEAR<br>PROTEIN) - HOMO SAPIENS<br>(HUMAN), 249 aa. | 1.20E-89<br>9         |
| 311 | cg42937321 | 1977 | CTTTCCCTCTTAC<br>CCTCTCTCTCT(GT)<br>AACATCGTAACA<br>ACAGACTTACGT   | G<br>T |  | SILENT-<br>NONCOD<br>ING | potassium chan<br>nel | Human Gene SWISSPROT-<br>ID:P22001 VOLTAGE-GATED<br>POTASSIUM CHANNEL<br>PROTEIN KV1.3 (HPCN3)<br>(HGK5) (HUKII) (HLK3)-<br>HOMO SAPIENS (HUMAN), 523<br>aa.   | 5.40E-284<br>1 (1p21) |

|     |            |      |   |   |   |                   |                       |  |           |               |
|-----|------------|------|---|---|---|-------------------|-----------------------|--|-----------|---------------|
| 312 | cg42937321 | 1983 | CCCTCTTACCCCTCTC<br>TCTCTGAAACATCT<br>IGTAACACAACAGA<br>CTTACGTTAACT    | C | T | SILENT-<br>NONCOD | potassium Chan<br>nel | Human Gene SWISSPROT-<br>ID:P22001(VOLTAGE-GATED<br>POTASSIUM CHANNEL<br>PROTEIN KV1.3 (HPCN3)<br>(HGK5) (HUKII) (HLK3)-<br>HOMO SAPIENS (HUMAN), 523<br>aa.   | 5.40E-284 | 1 (1p21)      |
| 313 | cg40991963 | 1357 | CAAAATGTAAACAG<br>TGGCTTTCAACJA/<br>GJGGAGTAAAGCA<br>AAGTCTCTAAAGC<br>T | A | G | SILENT-<br>NONCOD | potassium Chan<br>nel | Human Gene SWISSPROT-<br>ID:P48048 ATP-SENSITIVE<br>INWARD RECTIFIER<br>POTASSIUM CHANNEL 1<br>(POTASSIUM CHANNEL,<br>INWARDLY RECTIFYING,<br>SUBFAMILY I, MEMBER 1)<br>(ATP-REGULATED)<br>POTASSIUM CHANNEL ROM-<br>K (KIR1.1) - HOMO SAPIENS<br>(HUMAN), 391 aa. | 1.80E-205 | 11<br>(11q24) |

|     |            |      |   |   |   |                           |               |  |   |               |
|-----|------------|------|---|---|---|---------------------------|---------------|--|---|---------------|
| 314 | cg43951366 | 2332 | AAAGATGTTGAA<br>TACTTAAACACTG/<br>AATCACAAAGATGGC<br>AAAATGCTGAAG | G | A | SILENT-<br>NONCOD-<br>ING | prostaglandin | Human Gene SWISSNEW<br>ID:P35354 PROSTAGLANDIN<br>G/H SYNTHASE 2 PRECURSOR<br>(EC 1.14.99.1)<br>(CYCLOOXYGENASE -2) (COX-<br>2) (PROSTAGLANDIN-<br>ENDOPEROXIDE SYNTHASE 2)<br>(PROSTAGLANDIN H2<br>SYNTHASE 2) (PGH SYNTHASE<br>2) (PGHS-2) (PGS II) - HOMO<br>SAPIENS (HUMAN), 604<br>aa.[pcl:SPTREMBL;id:Q16876<br>PROSTAGLANDIN<br>ENDOPEROXIDE SYNTHASE-2<br>PRECURSOR (EC 1.14.99.1)<br>(PROSTAGLANDIN-<br>ENDOPEROXIDE SYNTHASE)<br>(PROSTAGLANDIN<br>SYNTHASE)<br>(PROSTAGLANDIN G/H<br>SYNTHASE) - HOMO SAPIENS<br>(HUMAN), 604 aa. | 0 | 1<br>(1625.2) |
|-----|------------|------|---|---|---|---------------------------|---------------|--|---|---------------|

|     |            |      |   |   |   |                           |               |   |                       |               |
|-----|------------|------|---|---|---|---------------------------|---------------|---|-----------------------|---------------|
| 315 | cgt3951366 | 2829 | TGGTGGAGCCACT<br>GCAGTGTATCTT/<br>CIAAAATAAGAAAT<br>ATTTGTTGAGATA | T | C | SILENT-<br>NONCOD-<br>ING | prostaglandin | Human Gene SWISSNEW-<br>ID:P35354 PROSTAGLANDIN<br>G/H SYNTHASE 2 PRECURSOR<br>(EC 1.14.99.1)<br>(CYCLOOXYGENASE -2) (COX-<br>2) (PROSTAGLANDIN H2<br>SYNTHASE 2) (PGH SYNTHASE<br>2) (PGHS-2) (PHS II) - HOMO<br>SAPIENS (HUMAN), 604<br>aa.[pcl:SPTRMBL-ID:Q16876<br>PROSTAGLANDIN<br>ENDOPEROXIDE SYNTHASE-2<br>PRECURSOR (EC 1.14.99.1)<br>(PROSTAGLANDIN H2<br>ENDOPEROXIDE SYNTHASE)<br>(PROSTAGLANDIN H2<br>SYNTHASE)<br>(PROSTAGLANDIN G/H<br>SYNTHASE) -HOMO SAPIENS<br>(HUMAN), 604 aa. | 0                     | 1<br>(1q25.2) |
| 316 | cgt3306254 | 1431 | CACTTAACCTTGCAT<br>GTGCACAGCTT/C<br>TGGTAACAAATAT<br>CGCTAAACCTTA | T | C | SILENT-<br>NONCOD-<br>ING | prostaglandin | Human Gene SPTRMBL-<br>ID:Q00325 PROSTAGLANDIN<br>EP3 RECEPTOR SUBTYPE<br>ISOFORM - HOMO SAPIENS<br>(HUMAN), 402 aa.  | 1.40E-211<br>(1p31.2) |               |

|     |            |      |   |   |   |  |                           |               |   |                            |  |
|-----|------------|------|---|---|---|--|---------------------------|---------------|---|----------------------------|--|
| 317 | cg43306254 | 1666 | AATGTGATTAAATTAT<br>GTGATGAAAAC/A/<br>TTTTTATAAAAT<br>GATCTTGGTCTAT | A | T |  | SILENT-<br>NONCOD-<br>ING | prostaglandin | Human Gene SPTREMBL-<br>ID:O00325 PROSTAGLANDIN<br>EP3 RECEPTOR SUBTYPE<br>ISOFORM - HOMO SAPIENS<br>(HUMAN), 402 aa.   | 1.40E-211<br>1<br>(Ip31.2) |  |
| 318 | cg42918089 | 1064 | CAATCAGAAATTGA<br>TAAGGCACTGTC[C/<br>T]TCCACTCCATT<br>AGCAAATTAGTCA | C | T |  | SILENT-<br>NONCOD-<br>ING | protease      | Human Gene Homologous to<br>SWISSPROT-ID:P09237<br>MATRIX LYNSIN PRECURSOR (EC<br>3.4.24.23) (PUMP-1 PROTEASE)<br>(UTERINE<br>METALLOPROTEINASE)<br>(MATRIX<br>METALLOPROTEINASE-7)<br>(MMP-7) (MATRIN) - HOMO<br>SAPIENS (HUMAN), 267<br>aa. pcis:SWISSPROT-ID:P09237<br>MATRIX LYNSIN PRECURSOR (EC<br>3.4.24.23) (PUMP-1 PROTEASE)<br>(UTERINE<br>METALLOPROTEINASE)<br>(MATRIX<br>METALLOPROTEINASE-7)<br>(MMP-7) (MATRIN) - HOMO<br>SAPIENS (HUMAN), 267 aa. | 2.40E-146<br>11<br>(11q21) |  |
| 319 | cg44032168 | 1703 | TCCATCCCTCTTT<br>GGGCCTCTCTG/C<br>AAGGAAGTAACA<br>TTTACTGAGCACC     | G | C |  | SILENT-<br>NONCOD-<br>ING | protease      | Human Gene Similar to<br>SWISSPROT-ID:P25155<br>COAGULATION FACTOR X<br>PRECURSOR (EC 3.4.21.6)<br>(STUART FACTOR) (VIRUS<br>ACTIVATING PROTEASE)<br>(VAP) - GALLUS GALLUS<br>(CHICKEN), 475 aa.  | 2.40E-82<br>2<br>(2q13)    |  |

|     |            |      |   |     |   |                          |           |  |           |               |
|-----|------------|------|---|-----|---|--------------------------|-----------|--|-----------|---------------|
| 320 | cg43154190 | 1250 | TACCCGGAAAGTTG<br>AGCTCAATTCA[IV]<br>CJTCTGTTCCTGGA<br>CCACAACTGCCA       | T   | C | SILENT-<br>NONCOD<br>ING | protease  | Human Gene Similar to<br>SWISSPROT-ID:P50280<br>MATRILYSIN PRECURSOR (EC<br>3.4.24.23) (PUMP-1 PROTEASE)<br>(UTERINE<br>METALLOPROTEINASE)<br>(MATRIX<br>METALLOPROTEINASE-7)<br>(MMP-7) (MATRIN) - RATIUS<br>NORVEGICUS (RAT), 267 aa.                        | 2.40E-59  | 11<br>(11q22) |
| 321 | cg43927549 | 175  | CCCAGTCCCTGCCG<br>CTCCTACTGGGG[A<br>/C]GTGCCGCTGGTC<br>GGAAAGATGCTGG<br>A | A   | C | SILENT-<br>NONCOD<br>ING | reductase | Human Gene Homologous to<br>SWISSPROT-ID:P16083<br>NAD(PH) DEHYDROGENASE<br>(QUINONE) 2 (EC 1.6.99.2)<br>(QUINONE REDUCTASE) (DT-<br>DIAPHORASE)<br>(AZOREDUCTASE)<br>(PHYLLOQUINONE<br>REDUCTASE) (MENADIONE<br>REDUCTASE) - HOMO<br>SAPIENS (HUMAN), 231 aa. | 1.60E-124 | 6 (6pter)     |
| 322 | cg43927549 | 191  | TACTGGGGAGTGC<br>GCTGGGTGGAAG[<br>ap/G]ATTGCTGGAC<br>TCGCTGAAGAGAG<br>AC  | gap | G | SILENT-<br>NONCOD<br>ING | reductase | Human Gene Homologous to<br>SWISSPROT-ID:P16083<br>NAD(PH) DEHYDROGENASE<br>(QUINONE) 2 (EC 1.6.99.2)<br>(QUINONE REDUCTASE) (DT-<br>DIAPHORASE)<br>(AZOREDUCTASE)<br>(PHYLLOQUINONE<br>REDUCTASE) (MENADIONE<br>REDUCTASE) - HOMO<br>SAPIENS (HUMAN), 231 aa. | 1.60E-124 | 6 (6pter)     |

|     |            |      |   |     |     |  |                           |           |  |           |          |
|-----|------------|------|---|-----|-----|--|---------------------------|-----------|--|-----------|----------|
| 323 | cg43927549 | 52   | CGGTCCGGTCC<br>CGGGGGGGCAGI <sup>b</sup><br>ap/GTICGCAAGCGCT<br>CCCGCCCTCCAGG<br>CG | gap | G   |  | SILENT-<br>NONCOD-<br>ING | reductase | Human Gene Homologous to<br>SWISSPROT-ID:PI6083<br>NAD(P)H DEHYDROGENASE<br>(QUINONE) 2 (EC 1.6.99.2)<br>(QUINONE REDUCTASE) (DT-<br>DIAPHORASE)<br>(AZOREDUCTASE)<br>(PHYLLOQUINONE<br>REDUCTASE) (MENADIONE<br>REDUCTASE) - HOMO<br>SAPIENS (HUMAN), 231 aa. | 1.60E-124 | 6 (6per) |
| 324 | cg43947066 | 780  | TTCCTAAAGGCT<br>GGGGCTATTATA<br>/GTTAAGAACATT<br>CCAAAGTGACTCT                      | A   | G   |  | SILENT-<br>NONCOD-<br>ING | strict    | Human Gene SWISSPROT-<br>ID:O15142 ACTIN-LIKE<br>PROTEIN 2 - HOMO SAPIENS<br>(HUMAN), 394 aa.  | 3.30E-207 | 2        |
| 325 | cg43923264 | 113  | AGGAAGCCGGAG<br>AATTGGGGCACGIC<br>/gapIAAGAGGGGG<br>GCCTTGATGACCC<br>GC             | C   | gap |  | SILENT-<br>NONCOD-<br>ING | strict    | Human Gene SWISSPROT-<br>ID:Q14012<br>CALCIUM/CALMODULIN-<br>DEPENDENT PROTEIN KINASE<br>TYPE I (EC 2.7.1.123) (CAM<br>KINASE I) - HOMO SAPIENS<br>(HUMAN), 370 aa.  | 1.70E-200 | 3        |
| 326 | cg43942332 | 1926 | AGATTCAATCAGAA<br>TAGGATTTTGCCIA/<br>CIAAACTCCACCCA<br>TATGCTGTGAGC                 | A   | C   |  | SILENT-<br>NONCOD-<br>ING | strict    | Human Gene Homologous to<br>SPTREMBL-ID:Q00379 DELTA-<br>CATENIN - HOMO SAPIENS<br>(HUMAN), 792 aa.  | 2.10E-124 | 11       |

|     |             |     |  |   |   |                           |        |  |           |          |
|-----|-------------|-----|--|---|---|---------------------------|--------|--|-----------|----------|
| 327 | cgi3274705  | 580 | CCGCTGTCTCTGTC<br>TTCGCTTTA[T/G/T]<br>TCAAGAAAGAATAA<br>TGCGACGAAAAT | G | T | SILENT-<br>NONCOD-<br>ING | struct | Human Gene Homologous to<br>SPTREMBL-ID:Q28910 MUCIN -<br>BOS TAURUS (BOVINE), 600 aa<br>(fragment).   | 4.80E-110 | 12       |
| 328 | cgi42207316 | 146 | CCACTCTCTGGGA<br>CACATTGCCCTTC[T<br>TGTTTTCTCCAGC<br>ATGGCTTGCTGCIC  | C | T | SILENT-<br>NONCOD-<br>ING | struct | Human Gene Similar to<br>SWISSPROT-ID:P12273<br>PROLACTIN-INDUCIBLE<br>PROTEIN PRECURSOR<br>(SECRETORY ACTIN-BINDING<br>PROTEIN) (SABP) (GROSS<br>CYSTIC DISEASE FLUID<br>PROTEIN 15) (GCDFP-15)<br>(GP17)-HOMO SAPIENS<br>(HUMAN), 146<br>aa. pcds SWISSPROT-ID:P12273<br>PROLACTIN-INDUCIBLE<br>PROTEIN PRECURSOR<br>(SECRETORY ACTIN-BINDING<br>PROTEIN) (SABP) (GROSS<br>CYSTIC DISEASE FLUID<br>PROTEIN 15) (GCDFP-15)-<br>HOMO SAPIENS (HUMAN), 146<br>aa. | 3.50E-74  | 7 (7q32) |
| 329 | cgi43927885 | 546 | CATCATCATCATA<br>GTTTACTTCAGCQA/<br>TCTTAAATCCCCG<br>AGGAGTCGCCCT    | A | T | SILENT-<br>NONCOD-<br>ING | struct | Human Gene Similar to<br>SWISSPROT-ID:P19065<br>SYNAPTOBREVIN 2 (VESICLE<br>ASSOCIATED MEMBRANE<br>PROTEIN 2) (VAMP-2) - HOMO<br>SAPIENS (HUMAN), AND BOS<br>TAURUS (BOVINE), 115 aa.  | 1.20E-55  | 17       |

|     |            |      |   |   |   |                          |          |   |           |                     |
|-----|------------|------|---|---|---|--------------------------|----------|---|-----------|---------------------|
| 330 | cg40388639 | 5029 | CTCTTGCCCAGGCC<br>GCTGCAAGTTTgap<br>TGTAAAGCGGG<br>ACAGACACTGCTG<br>A | T |   | SILENT-<br>NONCOD<br>ING | synthase | Human Gene SWISSPROT-<br>ID:P29475 NITRIC-OXIDE<br>SYNTHASE, BRAIN (EC<br>1.14.13.39) (NOS, TYPE I)<br>(NEURONAL NOS) (NNOS) -<br>HOMO SAPIENS (HUMAN),<br>1434 aa.   | 0         | 12<br>(12q24.2<br>) |
| 331 | cg43949316 | 555  | AGGTACCAAACA<br>GGAATACAAACAC[C<br>T]TCTCTCCCTTT<br>CTGCTCTAGAAGG     | C | T | SILENT-<br>NONCOD<br>ING | synthase | Human Gene SWISSPROT-<br>ID:P48651<br>PHOSPHATIDYL SERINE<br>SYNTHASE I (SERINE-<br>EXCHANGE ENZYME I) (EC<br>2.7.8.-) (Q1AA0024) - HOMO<br>SAPIENS (HUMAN), 473 aa.  | 9.80E-269 | 8                   |
| 332 | cg43958714 | 1565 | TGGGTGATGATCA<br>CTGGCTGCTGT[G/<br>C]GGCTCATGGCAG<br>AGCATTCAAGTGCC   | T | C | SILENT-<br>NONCOD<br>ING | synthase | Human Gene Similar to<br>SPTREM1BL-1D:Q42761<br>SQUALENE SYNTHASE (EC<br>2.5.1.21) (FARNESYL-<br>DIPHOSPHATE<br>FARNESYLTRANSFERASE)<br>(FARNESYLTRANSFERASE)<br>(PRESQUALENE-DI-<br>DIPHOSPHATE<br>SYNTHASE) - GLYCYYRRHIZA<br>GLABRA, 412 aa. | 9.20E-83  | 8                   |

|     |            |      |  |   |   |                           |          |   |          |          |
|-----|------------|------|--|---|---|---------------------------|----------|---|----------|----------|
| 333 | cg43275028 | 2508 | ACAGACTGGCTGC<br>AGCATTAGGAATIC<br>TJAGGTCAATTCCGA<br>AACTCATATTGA       | C | T | SILENT-<br>NONCOD-<br>ING | synthase | Human Gene Similar to<br>SWISSPROT-ID:P70490 MILK<br>FAT GLOBULE-EGF FACTOR 8<br>PRECURSOR (MFG-E8) (O-<br>ACETYL GD3 GANGLIOSIDE<br>SYNTHASE) (AGS) (MFGM)<br>RATTUS NORVEGICUS (RAT),<br>427 aa. pcis:SPTREM/BL-<br>ID:P70490 O-ACETYL GD3<br>GANGLIOSIDE SYNTHASE -<br>RATTUS NORVEGICUS (RAT),<br>427 aa.   | 3.20E-65 | 1 (1q23) |
| 334 | cg43275028 | 2535 | GGTCATTCCGAAA<br>CTCATCATTTGAAT/<br>C]CAGGAAAGAAGA<br>AGAGTTCAATCTT<br>A | T | C | SILENT-<br>NONCOD-<br>ING | synthase | Human Gene Similar to<br>SWISSPROT-ID:P70490 MILK<br>FAT GLOBULE-EGF FACTOR 8<br>PRECURSOR (MFG-E8) (O-<br>ACETYL GD3 GANGLIOSIDE<br>SYNTHASE) (AGS) (MFGM) -<br>RATTUS NORVEGICUS (RAT),<br>427 aa. pcis:SPTREM/BL-<br>ID:P70490 O-ACETYL GD3<br>GANGLIOSIDE SYNTHASE -<br>RATTUS NORVEGICUS (RAT),<br>427 aa. | 3.20E-65 | 1 (1q23) |

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| 335 | cg43275028 | 2601 | AGAATGGCACTGA<br>ATTCGTTCTTCIA/<br>GAAACACAGATAA<br>ATTGTTGGTCAA       | A | G | SILENT-<br>NONCOD-<br>ING | synthase | Human Gene Similar to<br>SWISSPROT-ID:P70490 MILK<br>FAT GLOBULE-EGF FACTOR 8<br>PRECURSOR (MFG-E8) (O-<br>ACETYL GD3 GANGLIOSIDE<br>SYNTHASE) (AGS) (MFGM)-<br>RATTUS NORVEGICUS (RAT),<br>427 aa.[pcis:SPTREMBL-<br>ID:P70490 O-ACETYL GD3<br>GANGLIOSIDE SYNTHASE -<br>RATTUS NORVEGICUS (RAT),<br>427 aa. | 3.20E-65 | 1 (1q23) |
| 336 | cg43275028 | 2873 | CTTCACTGGTGC<br>TGGAGTAATTCAIA/<br>GIAAGTCAAGAAC<br>ATGCTAAGCATAA<br>G | A | G | SILENT-<br>NONCOD-<br>ING | synthase | Human Gene Similar to<br>SWISSPROT-ID:P70490 MILK<br>FAT GLOBULE-EGF FACTOR 8<br>PRECURSOR (MFG-E8) (O-<br>ACETYL GD3 GANGLIOSIDE<br>SYNTHASE) (AGS) (MFGM)-<br>RATTUS NORVEGICUS (RAT),<br>427 aa.[pcis:SPTREMBL-<br>ID:P70490 O-ACETYL GD3<br>GANGLIOSIDE SYNTHASE -<br>RATTUS NORVEGICUS (RAT),<br>427 aa. | 3.20E-65 | 1 (1q23) |

|     |            |      |  |        |                          |          |   |          |          |
|-----|------------|------|--|--------|--------------------------|----------|---|----------|----------|
| 337 | cgt3275028 | 2894 | TTCAAAAGTCAAG<br>AACATGCTAACGCA<br>GGTAAGGGACCCA<br>AGGTAGAAAGAGA<br>T | A<br>G | SILENT-<br>NONCOD<br>ING | synthase | Human Gene Similar to<br>SWISSPROT-ID:P70490 MILK<br>FAT GLOBULE-EGF FACTOR 8<br>PRECURSOR (MFG-E8) (O-<br>ACETYL GD3 GLANGLIOSIDE<br>SYNTHASE) (AGS) (MFGM)-<br>RATTUS NORVEGICUS (RAT),<br>427 aa.[pcis:SPTREMBL,<br>ID:P70490 O-ACETYL GD3<br>GLANGLIOSIDE SYNTHASE -<br>RATTUS NORVEGICUS (RAT),<br>427 aa. | 3.20E-65 | 1 (1q23) |
| 338 | cgt3275028 | 3073 | TTCCTCCAGAA<br>TGAGGGCCCTGGIA/<br>GJAGGACCCCTCCCTA<br>GTGATCTTTACT     | A<br>G | SILENT-<br>NONCOD<br>ING | synthase | Human Gene Similar to<br>SWISSPROT-ID:P70490 MILK<br>FAT GLOBULE-EGF FACTOR 8<br>PRECURSOR (MFG-E8) (O-<br>ACETYL GD3 GLANGLIOSIDE<br>SYNTHASE) (AGS) (MFGM)-<br>RATTUS NORVEGICUS (RAT),<br>427 aa.[pcis:SPTREMBL,<br>ID:P70490 O-ACETYL GD3<br>GLANGLIOSIDE SYNTHASE -<br>RATTUS NORVEGICUS (RAT),<br>427 aa. | 3.20E-65 | 1 (1q23) |

|     |            |      |  |   |   |                          |          |  |           |               |
|-----|------------|------|--|---|---|--------------------------|----------|--|-----------|---------------|
| 339 | cg43275028 | 5590 | ACTACATAAGGAC<br>AGCAACATGCCCTA<br>/GTGGACATGAGA<br>GAATTGGTCTACT  | A | G | SILENT-<br>NONCOD<br>ING | Synthase | Human Gene Similar to<br>SWISSPROT-ID:P70490 MILK<br>FAT GLOBULE-EGF FACTOR 8<br>PRECURSOR (MFG-E8) (O-<br>ACETYL GD3 GANGLIOSIDE<br>SYNTHASE) (AGS) (MFGM)-<br>RATTUS NORVEGICUS (RAT),<br>427 aa.[pcis:SPTREM(BL-<br>ID:P70490 O-ACETYL GD3<br>GANGLIOSIDE SYNTHASE -<br>RATTUS NORVEGICUS (RAT),<br>427 aa. | 3.20E-65  | 1 (1q23)      |
| 340 | cg43985000 | 1856 | GAAAAAAATCACA<br>AGGCAACTGTGAIC<br>/GTCGGGGAAATCT<br>CTTCCTGATCCIT | C | G | SILENT-<br>NONCOD<br>ING | tm7      | Human Gene SWISSPROT-<br>ID:P25101 ENDOTHELIN-1<br>RECEPTOR PRECURSOR (ET-A)<br>-HOMO SAPIENS (HUMAN),<br>427 aa.  | 1.60E-236 | 4             |
| 341 | cg39565524 | 1684 | TCCGACCCACAC<br>ACCCTGAGGGAGC<br>/GJCCCTACCTAGCC<br>TCAGCCGCTCCTG  | C | G | SILENT-<br>NONCOD<br>ING | tm7      | Human Gene SWISSPROT-<br>ID:P51575 P2X PURINOCEPTOR<br>1 (ATP RECEPTOR) (P2X1)<br>(PURINERGIC RECEPTOR)-<br>HOMO SAPIENS (HUMAN), 399<br>aa.   | 2.00E-220 | 17            |
| 342 | cg43306266 | 1603 | ATAATCCATGCCCT<br>TGAATATTAGATV<br>GTGGTTCTTGGA<br>TGGGATTGTGAAT   | T | G | SILENT-<br>NONCOD<br>ING | tm7      | Human Gene SWISSPROT-<br>ID:P43115 PROSTAGLANDIN E2<br>RECEPTOR, EP3 SUBTYPE<br>(PROSTANOID EP3 RECEPTOR)<br>(PGE RECEPTOR, EP3<br>SUBTYPE) - HOMO SAPIENS<br>(HUMAN), 390 aa.   | 4.80E-212 | 1<br>(1p31.2) |

|     |            |      |   |     |   |  |                           |     |  |           |               |
|-----|------------|------|---|-----|---|--|---------------------------|-----|--|-----------|---------------|
| 343 | cg43306266 | 1641 | GGGATTTCATA<br>TGCATTTAGAAAG<br>ap/CGTGGAAAGA<br>ATTCACAGATGA<br>TG   | gap | C |  | SILENT-<br>NONCOD-<br>ING | tm7 | Human Gene SWISSPROT-<br>ID:P43115 PROSTAGLANDIN E2<br>RECEPTOR, EP3 SUBTYPE<br>(PROSTANOID EP3 RECEPTOR)<br>(PGE RECEPTOR, EP3<br>SUBTYPE) - HOMO SAPIENS<br>(HUMAN), 390 aa. | 4.80E-212 | 1<br>(1p31.2) |
| 344 | cg43306266 | 1650 | GAATATGCCATT<br>AGAACGTTGGAA[<br>GIC]AATTACACAG<br>ATGATGATGGAG<br>GA | G   | C |  | SILENT-<br>NONCOD-<br>ING | tm7 | Human Gene SWISSPROT-<br>ID:P43115 PROSTAGLANDIN E2<br>RECEPTOR, EP3 SUBTYPE<br>(PROSTANOID EP3 RECEPTOR)<br>(PGE RECEPTOR, EP3<br>SUBTYPE) - HOMO SAPIENS<br>(HUMAN), 390 aa. | 4.80E-212 | 1<br>(1p31.2) |

|     |            |     |   |     |   |                           |     |   |           |   |
|-----|------------|-----|---|-----|---|---------------------------|-----|---|-----------|---|
| 345 | cg43329467 | 683 | TCGGCAAATCTTG<br>AAAGCTGCAGGGT<br>CTTGCGAGAGACAT<br>GGATGTGACTTCC<br>CA | C   | T | SILENT-<br>NONCOD-<br>ING | tm7 | Human Gene SWISSNEW-<br>ID:Q99527 CHEMOKINE<br>RECEPTOR-LIKE 2 (IL8-<br>RELATED RECEPTOR DRY12)<br>(FLOW-INDUCED<br>ENDOTHELIAL G PROTEIN-<br>COUPLED RECEPTOR) (FEG-1)<br>(G PROTEIN-COUPLED<br>RECEPTOR GPR30) (GPCR-BR)<br>- HOMO SAPIENS (HUMAN),<br>375 aa. pcis:SWISSPROT-<br>ID:Q99527 CHEMOKINE<br>RECEPTOR-LIKE 2 (IL8-<br>RELATED RECEPTOR DRY12)<br>(FLOW-INDUCED<br>ENDOTHELIAL G PROTEIN-<br>COUPLED RECEPTOR) (FEG-1)<br>(G PROTEIN-COUPLED<br>RECEPTOR GPR30) - HOMO<br>SAPIENS (HUMAN), 375<br>aa. pcis:TREMBLNW-<br>ID:G2656121 G-PROTEIN<br>COUPLED RECEPTOR - HOMO<br>SAPIENS (HUMAN), 375 aa. | 8.20E-201 | 7 |
| 346 | cg2751286  | 439 | AAGGCATAAGAAC<br>TAGGAGCTGCTG <br>ap/GIACATTCAAT<br>ATGAAGGGCAACT<br>CC | gap | G | SILENT-<br>NONCOD-<br>ING | tm7 | Human Gene SWISSPROT-<br>ID:P50052 TYPE-2<br>ANGIOTENSIN II RECEPTOR<br>(AT2) - HOMO SAPIENS<br>(HUMAN), 363 aa.  | 2.00E-197 | X |

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| 347 | cg11751407 | 76   | GAATGTGGGATA<br>AGGCATTGGAC[C<br>T]CCTATCAGGTATC<br>CTGAGGAGACT    | C | T |  | SILENT-<br>NONCOD-<br>ING | tm7 | Human Gene SWISSPROT-<br>ID:P46089 PROBABLE G<br>PROTEIN-COUPLED<br>RECEPTOR GPR3 (ACCA<br>ORPHAN RECEPTOR) - HOMO<br>SAPIENS (HUMAN), 330 aa. | 3.20E-176 | 1       |
| 348 | cg43326635 | 135  | CAGCCGGAGCTC<br>TGCCAGCTTGG[G/C<br>T]GAAGGGGGTG<br>CTTGCCTCGTCCC   | C | T |  | SILENT-<br>NONCOD-<br>ING | tm7 | Human Gene SWISSPROT-<br>ID:P30542 ADENOSINE A1<br>RECEPTOR - HOMO SAPIENS<br>(HUMAN), 326 aa.   | 1.10E-173 | 1       |
| 349 | cg43326635 | 139  | CGGGAGCTCTGCC<br>AGCTTTGGGAA[G<br>/C]GAGGGTGCTTG<br>CCTCGTCCCCCTTG | G | C |  | SILENT-<br>NONCOD-<br>ING | tm7 | Human Gene SWISSPROT-<br>ID:P30542 ADENOSINE A1<br>RECEPTOR - HOMO SAPIENS<br>(HUMAN), 326 aa.   | 1.10E-173 | 1       |
| 350 | cg43993798 | 1839 | TGCTCTTGCTGTG<br>ATGGAGGAGGA[A<br>G]GGGGTGATCCC<br>GTGGAGGCCCAA    | A | G |  | SILENT-<br>NONCOD-<br>ING | tm7 | Human Gene Homologous to<br>SWISSPROT-ID:P31421<br>METABOTROPIC GLUTAMATE<br>RECEPTOR 2 PRECURSOR -<br>RATTUS NORVEGICUS (RAT),<br>872 aa.     | 6.90E-109 | 3 (342) |

|     |            |      |   |   |   |  |                           |     |  |          |
|-----|------------|------|---|---|---|--|---------------------------|-----|--|----------|
| 351 | cg43040271 | 2130 | ATGCTTCCCCAAC<br>CCTAGGGAAATC/A/<br>CIAACACTTAAGATA<br>ATTGCCACCTCT | A | C |  | SILENT-<br>NONCOD-<br>ING | tm7 | Human Gene Similar to<br>SWISSPROT-ID:Q25322<br>TYRAMINE/OCTOPAMINE<br>RECEPTOR 2 (TYR-LOC 2) -<br>LOCUSTA MIGRATORIA<br>(MIGRATORY LOCUST), 484<br>aa.Ipcis:SPTREMBL-ID:Q25322<br>GCR2 (G PROTEIN-COUPLED<br>RECEPTOR) • LOCUSTA<br>MIGRATORIA (MIGRATORY<br>LOCUST), 484 aa. | 2.90E-74 |
| 352 | cg43040271 | 2139 | CCAACCCTAGGGA<br>ATCAAACACTTAA/G<br>TATAATTGGCCAC<br>TTCTCCCTTTCT   | G | T |  | SILENT-<br>NONCOD-<br>ING | tm7 | Human Gene Similar to<br>SWISSPROT-ID:Q25322<br>TYRAMINE/OCTOPAMINE<br>RECEPTOR 2 (TYR-LOC 2) -<br>LOCUSTA MIGRATORIA<br>(MIGRATORY LOCUST), 484<br>aa.Ipcis:SPTREMBL-ID:Q25322<br>GCR2 (G PROTEIN-COUPLED<br>RECEPTOR) • LOCUSTA<br>MIGRATORIA (MIGRATORY<br>LOCUST), 484 aa. | 2.90E-74 |

|     |            |      |  |        |  |                          |     |   |          |
|-----|------------|------|--|--------|--|--------------------------|-----|---|----------|
| 353 | cg43040271 | 2163 | AGATAATTCGCCA<br>CTTCTCCCTTC/<br>TTCTCTGCTCCGC<br>TCACGGCTGCAG     | C<br>T |  | SILENT-<br>NONCOD<br>ING | tm7 | Human Gene Similar to<br>SWISSPROT-ID:Q25322<br>TYRAMINE/OCTOPAMINE<br>RECEPTOR 2 (TYR-LOC 2)-<br>LOCUSTA MIGRATORIA<br>(MIGRATORY LOCUST), 484<br>aa[publ:SPTREMBL-ID:Q25322<br>GCR2 (G PROTEIN-COUPLED<br>RECEPTOR) -LOCUSTA<br>MIGRATORIA (MIGRATORY<br>LOCUST), 484 aa. | 2.90E-74 |
| 354 | cg43040273 | 1668 | CGCAAGCCCCGC<br>CGTGGGTCCGCC/T<br>CJGCTGAGGCC<br>CCAGCCAGTGCGC     | T<br>C |  | SILENT-<br>NONCOD<br>ING | tm7 | Human Gene Similar to<br>SWISSPROT-ID:Q24563<br>DOPAMINE RECEPTOR 2 -<br>DROSOPHILA<br>MELANOGLASTER (FRUIT FLY),<br>539 aa.  | 2.00E-58 |
| 355 | cg43040273 | 1760 | CAGGCCCTCTTGC<br>TGGCACCCATA/<br>GGAAGCCATGCC<br>CGGACCAAGACGT     | A<br>G |  | SILENT-<br>NONCOD<br>ING | tm7 | Human Gene Similar to<br>SWISSPROT-ID:Q24563<br>DOPAMINE RECEPTOR 2 -<br>DROSOPHILA<br>MELANOGLASTER (FRUIT FLY),<br>539 aa.  | 2.00E-58 |
| 356 | cg43040273 | 1793 | TGGCCGGACCAC<br>GACGTCAAGCAGIC<br>/GAAAGGGACGAG<br>GTGTGGGTGG<br>G | C<br>G |  | SILENT-<br>NONCOD<br>ING | tm7 | Human Gene Similar to<br>SWISSPROT-ID:Q24563<br>DOPAMINE RECEPTOR 2 -<br>DROSOPHILA<br>MELANOGLASTER (FRUIT FLY),<br>539 aa.  | 2.00E-58 |

|     |            |      |   |     |   |  |                           |                 |  |           |          |
|-----|------------|------|---|-----|---|--|---------------------------|-----------------|--|-----------|----------|
| 357 | cg43040273 | 2767 | GCAGGGTCTTCCTTG<br>AAGGCCATATGG[G/<br>C]AATGGCTACTCC<br>AGCAACGGCAACA       | G   | C |  | SILENT-<br>NONCOD-<br>ING | tm7             | Human Gene Similar to<br>SWISSPROT-ID:Q24563<br>DOPAMINE RECEPTOR 2 -<br>DROSOPHILA<br>MELANOOGASTER (FRUIT FLY),<br>539 aa. | 2.00E-58  | 5 (5q32) |
| 358 | cg43040273 | 2953 | ATTGTAGTACAAA<br>TGACTCACTGCTG[<br>A]TAAGGAGTTT<br>TCTACTTTAAAG             | G   | A |  | SILENT-<br>NONCOD-<br>ING | tm7             | Human Gene Similar to<br>SWISSPROT-ID:Q24563<br>DOPAMINE RECEPTOR 2 -<br>DROSOPHILA<br>MELANOOGASTER (FRUIT FLY),<br>539 aa. | 2.00E-58  | 5 (5q32) |
| 359 | cg43040273 | 3053 | ATAAAACTTAGAAT<br>AAAATTGAAAA[<br>G]ap[A]TTGTATAGAG<br>ATAIGCAGAAAGGA<br>AG | gap | A |  | SILENT-<br>NONCOD-<br>ING | tm7             | Human Gene Similar to<br>SWISSPROT-ID:Q24563<br>DOPAMINE RECEPTOR 2 -<br>DROSOPHILA<br>MELANOOGASTER (FRUIT FLY),<br>539 aa. | 2.00E-58  | 5 (5q32) |
| 360 | cg43998970 | 1501 | AGGGGTGAACTG<br>CTGATGGATT[T]ga<br>p[T]CCTTCATTCCC<br>TCTGTATAAAGGT<br>A    | gap | T |  | SILENT-<br>NONCOD-<br>ING | transcripfactor | Human Gene SPTREMBL-<br>ID:Q07279 TRANSCRIPTION<br>FACTOR NF-E2 - MUS<br>MUSCULUS (MOUSE), 373 aa.                           | 1.70E-177 | 12       |
| 361 | cg43998970 | 249  | AGCCTCCCCAGAG<br>ACAAACACCGGGGA[<br>G]CJCCTCATCTCTC<br>TCCTCACCCCTGCTG      | G   | C |  | SILENT-<br>NONCOD-<br>ING | transcripfactor | Human Gene SPTREMBL-<br>ID:Q07279 TRANSCRIPTION<br>FACTOR NF-E2 - MUS<br>MUSCULUS (MOUSE), 373 aa.                           | 1.70E-177 | 12       |

|     |            |      |  |     |   |                           |                   |   |           |               |
|-----|------------|------|--|-----|---|---------------------------|-------------------|---|-----------|---------------|
| 362 | cg43947199 | 2623 | GTCTTCTCCGGGCC<br>CACCCCGCTGGC/T<br>[AAGGGGAAGTGG<br>GCGAAGCTGGAGC                 | C   | T | SILENT-<br>NONCOD-<br>ING | transcriptifactor | Human Gene SWISSNEW-<br>ID:P23193 TRANSCRIPTION<br>ELONGATION FACTOR S-II<br>(TRANSCRIPTION<br>ELONGATION FACTOR A)-<br>HOMO SAPIENS (HUMAN), 301<br>aa.[pcds]SWISSPROT-ID:P23193<br>TRANSCRIPTION<br>ELONGATION FACTOR S-II<br>(TRANSCRIPTION<br>ELONGATION FACTOR A)-<br>HOMO SAPIENS (HUMAN), 301<br>aa. | 4.20E-158 | 8             |
| 363 | cg43917396 | 934  | GGGGCCGGCACT<br>GCCAGGAAGGGI<br>A/GICTCGGGAGA<br>GGGAGCGGGCGGC<br>TG               | A   | G | SILENT-<br>NONCOD-<br>ING | transcriptifactor | Human Gene Similar to<br>TREMBBLNEW-ID:G2920821<br>TRANSCRIPTION FACTOR T-<br>BOX 5 - HOMO SAPIENS<br>(HUMAN), 518 aa.  | 6.90E-63  |               |
| 364 | cg40351913 | 2030 | AGACGAAGAGCCC<br>AGGAAGTCATCC[C<br>C]GCAATGGAGA<br>GACACGAACAAAC<br>C              | T   | C | SILENT-<br>NONCOD-<br>ING | transport         | Human Gene SWISSPROT-<br>ID:Q011959 SODIUM-<br>DEPENDENT DOPAMINE<br>TRANSPORTER (DA<br>TRANSPORTER) (DAT) - HOMO<br>SAPIENS (HUMAN), 620 aa.   | 0         | 5<br>(Sp15.3) |
| 365 | cg43921289 | 237  | CCACGGCTGCCA<br>GGAGCAAGCCGA <sup>b</sup><br>ap/A]GAGCAGCCG<br>GCCGGCGCACTCC<br>GA | gap | A | SILENT-<br>NONCOD-<br>ING | UNCLASSIFI<br>ED  | Human Gene SWISSPROT-<br>ACC:P02545 LAMIN A (70 KD<br>LAMIN) - Homo sapiens (Human),<br>664 aa.   | 0         | 1             |

|     |            |      |  |   |   |                          |                  |   |   |               |
|-----|------------|------|--|---|---|--------------------------|------------------|---|---|---------------|
| 366 | cg43928515 | 3196 | AAACAAATAAAGCC<br>CTTTTACTGACIA/<br>GJATGCACCCAACC<br>TITTCAGCTGAAG    | A | G | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene SWISSPROT-<br>ACC:Q14687 HYPOTHETICAL<br>PROTEIN KIAA0182 - Homo<br>sapiens (Human), 1157 aa<br>(fragment).  | 0 | 16            |
| 367 | cg43955093 | 1309 | AGAGTCAAAAATC<br>CAAGTTGGATT[C/<br>G]TAAGCAGCCTTG<br>ACAGTAATCACTG     | C | G | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene SWIPEMBL-<br>ACC:Q16084 P130 - HOMO<br>SAPIENS (HUMAN), 1139 aa.   | 0 | 16            |
| 368 | cg43955093 | 1336 | AAGCAGCCTTGAC<br>AGTAATCACTG[A/<br>G]TGGTAGGGAAA<br>AAAAGACAGTGG<br>G  | A | G | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene SWIPEMBL-<br>ACC:Q16084 P130 - HOMO<br>SAPIENS (HUMAN), 1139 aa.   | 0 | 16            |
| 369 | cg43925474 | 2206 | AGGCAAAAGCTCA<br>CAGTAATGTAT[A/<br>C]CCAGAACAGGG<br>GCCTAAGTGAAGG<br>T | A | C | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene SWIPEMBL-<br>ACC:P42566 EPIDERMAL<br>GROWTH FACTOR RECEPTOR<br>SUBSTRATE SUBSTRATE 15<br>(PROTEIN EPS15) (AF-1P<br>PROTEIN) - Homo sapiens<br>(Human), 896 aa. | 0 | 1 (Ip32)      |
| 370 | cg44014437 | 4893 | CTGGCTCCCANCTTC<br>GCCAGCCCTCA[A/<br>G]GTACAACCTCC<br>GGGTGTAGTGGGC    | A | G | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene SWIPEMBL-<br>ACC:P53675 CLATHRIN<br>HEAVY CHAIN 2 (CLH-22) -<br>Homo sapiens (Human), 1640 aa.   | 0 | 17<br>(17q11) |

|     |            |      |   |   |   |  |                          |                  |  |   |                 |
|-----|------------|------|---|---|---|--|--------------------------|------------------|--|---|-----------------|
| 371 | cg4014448  | 5114 | CTGCTCCCAACTTC<br>GCCAGCCCTCCAA[T]<br>GT[G]IACAAC[TC]<br>GCGTAGTGGGC    | A | G |  | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene SWISSPROT-<br>ACC:P53675 CLATHRIN<br>HEAVY CHAIN 2 (CLH-22) -<br>Homo sapiens (Human), 1640 aa.   | 0 | 17<br>(17q11)   |
| 372 | cg43973129 | 2242 | CACTTCACTGAAA<br>GACACCATTATTC[C/<br>A]TACCCAAAGGCA<br>GAAACTAGAACTT    | C | A |  | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene SWISSPROT-<br>ACC:P05060 SECRETOGRAININ<br>1 PRECURSOR (SG1)<br>(CHROMOGRANIN B) - Homo<br>sapiens (Human), 677 aa.   | 0 | 20<br>(20pter)  |
| 373 | cg43950657 | 1939 | GATAAGCTCAAG<br>CITATTGGGATC[C/<br>T]CTGATCAAATTCT<br>TCTGATGTGTT       | C | T |  | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene SWISSNEW-<br>ACC:Q13009 T-LYMPHOMA<br>INVASION AND METASTASIS<br>INDUCING PROTEIN 1 (TIAM1<br>PROTEIN) - Homo sapiens<br>(Human), 1591 aa.  | 0 | 21<br>(21q22.1) |
| 374 | cg43956384 | 2416 | TACAGCCATCTGT<br>ACCTACTGGAGC[C/<br>T]GGCAGAAGGGAA<br>GTCCACTAGTCA<br>C | C | T |  | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene SWISSPROT-<br>ACC:P13866<br>SODIUM/GLUCOSE<br>COTRANSPORTER 1<br>(NA(+)/GLUCOSE<br>COTRANSPORTER 1) (HIGH<br>AFFINITY SODIUM-GLUCOSE<br>COTRANSPORTER) - Homo<br>sapiens (Human), 664 aa. | 0 | 22<br>(22q13.1) |

|     |            |      |  |        |  |                          |                 |  |           |               |
|-----|------------|------|--|--------|--|--------------------------|-----------------|--|-----------|---------------|
| 375 | cg43992229 | 101  | AGCAGTGCAGCCC<br>CGGCGCGGAGCA[<br>G/A]GGAGCTCGG<br>CCGGCGCCGGCG<br>CC  | G<br>A |  | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>E | Human Gene SWISSPROT-<br>ACC:P23352 KALLMANN<br>SYNDROME PROTEIN<br>PRECURSOR (ADHESION<br>MOLECULE-LIKE X-LINKED)<br>- Homo sapiens (Human), 680 aa.                              | 0         | X<br>(Xp22.3) |
| 376 | cg44932392 | 260  | GAGAAAAAGCATG<br>GTACCCAAACCGA[A<br>/T]TTTCCACTTTC<br>AGCAATACTTCAC    | A<br>T |  | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>E | Human Gene TREMBL NEW-<br>ACC:AAD23581 CULLIN 2-<br>HOMO SAPIENS (HUMAN), 745<br>aa.   | 0         |               |
| 377 | cg44932392 | 323  | TAAAGTTTAAGA<br>AATGTTCATAATG[A<br>/T]CATGAGCTTG<br>AATATCTCTAGGC<br>A | A<br>T |  | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>E | Human Gene TREMBL NEW-<br>ACC:AAD23581 CULLIN 2-<br>HOMO SAPIENS (HUMAN), 745<br>aa.   | 0         |               |
| 378 | cg43981656 | 1121 | AGCAAAGAAACAC<br>TGGCAGAATTCC[A<br>/T]GCATTGCAA<br>ATTCTAAGTTTGG       | A<br>T |  | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>E | Human Gene TREMBL NEW-<br>ACC:CAA08974 GUANINE<br>NUCLEOTIDE-EXCHANGE<br>FACTOR - HOMO SAPIENS<br>(HUMAN), 548 aa.   | 1.60E-292 | 10            |
| 379 | cg44910613 | 366  | AAATAAATGTTTC<br>ATAGTCATTACIT/A<br>JCCTTACAATGGGA<br>GTGCTAAAATTTC    | T<br>A |  | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>E | Human Gene SWISSPROT-<br>ACC:P38567 HYALURONIDASE<br>PRECURSOR (EC 3.2.1.35)<br>(SPERM SURFACE PROTEIN<br>PH-20) (SPERM ADHESION<br>MOLECULE 1) - Homo sapiens<br>(Human), 509 aa. | 1.20E-280 | 7             |

|     |            |      |   |   |     |                          |                  |  |           |               |
|-----|------------|------|---|---|-----|--------------------------|------------------|--|-----------|---------------|
| 380 | cg44035104 | 189  | AAGAAACTGATGT/CCTAAACCGTCTCAGCATGGCTGTA                 | T | C   | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene SWISSPROT-<br>ACC:P37287 N-<br>ACETYLGLUCOSAMINYL-<br>PHOSPHATIDYLINOSITOL<br>BIOSYNTHETIC PROTEIN<br>(GLCNAC-PI SYNTHESIS<br>PROTEIN)<br>(PHOSPHATIDYLINOSITOL<br>GLYCAN<br>COMPLEMENTATION CLASS<br>A) (PIG-A) - Homo sapiens<br>(Human), 484 aa. | 4.70E-261 | X<br>(Xp22.1) |
| 381 | cg43929959 | 1643 | CAATGCATGAATCTGTACCTCTGGIG/SapJAGGGCACTCACATGCCGCCCCAGC | G | gap | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene SPTREMBL-<br>ACC:P78505 DIABETES<br>MELLITUS TYPE I<br>AUTOANTIGEN (ISLET CELL<br>AUTOANTIGEN P69) - HOMO<br>SAPIENS (HUMAN), 483 aa.   | 2.10E-258 | 7             |
| 382 | cg43950250 | 1961 | TTGTCATGATTCTTGATGTTCTCT[ga]TAATGGAAAACTAAGAGATGGAAATT  | C | gap | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene SWISSPROT-<br>ACC:P11926 ORNITHINE<br>DECARBOXYLASE (EC<br>4.1.1.17) (ODC) - Homo sapiens<br>(Human), 461 aa.   | 7.00E-251 | 2<br>(2p25)   |
| 383 | cg43064090 | 129  | GCCGAGTCGGCTG/GTGGCGGACCC/A/TAGGGAGCAGCAGTAGGGAAAGTTG   | A | T   | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene SWISSPROT-<br>ACC:P32754-4-<br>HYDROXYPHENYL PYRUVATE<br>DIOXYGENASE (EC 1.13.11.27)<br>(4HPPD) (HPD) - Homo sapiens<br>(Human), 392 aa.  | 4.80E-213 |               |

|     |            |      |  |   |     |                          |                   |   |                        |
|-----|------------|------|--|---|-----|--------------------------|-------------------|---|------------------------|
| 384 | cg43064090 | 130  | CCGAGTCGGCTGG<br>TGGCGGACCAA<br>T]GGGGAGCAGCC<br>AGTAGGGAAAGTTG<br>G   | A | T   | SILENT-<br>NONCOD<br>ING | UNCLASSIFIIE<br>D | Human Gene SWISSPROT-<br>ACC:P32754 4-<br>HYDROXYPHENYL PYRUVATE<br>DIOXYGENASE (EC 1.13.11.27)<br>(4HPPD) (HPD) - Homo sapiens<br>(Human), 392 aa. | 4.80E-213              |
| 385 | cg43064090 | 157  | GGGAGGCCAGT<br>AGGGAAAGTTGGG<br>C/GGAGTCCAGA<br>ATCAGGGGGCGTG<br>GC    | C | G   | SILENT-<br>NONCOD<br>ING | UNCLASSIFIIE<br>D | Human Gene SWISSPROT-<br>ACC:P32754 4-<br>HYDROXYPHENYL PYRUVATE<br>DIOXYGENASE (EC 1.13.11.27)<br>(4HPPD) (HPD) - Homo sapiens<br>(Human), 392 aa. | 4.80E-213              |
| 386 | cg43064090 | 61   | TAATCGGGAGGGC<br>TGGAGCGAGGG<br>C/GGGCCCCGCC<br>AGGGGGCGTGGTCA<br>GT   | C | G   | SILENT-<br>NONCOD<br>ING | UNCLASSIFIIE<br>D | Human Gene SWISSPROT-<br>ACC:P32754 4-<br>HYDROXYPHENYL PYRUVATE<br>DIOXYGENASE (EC 1.13.11.27)<br>(4HPPD) (HPD) - Homo sapiens<br>(Human), 392 aa. | 4.80E-213              |
| 387 | cg30490224 | 3296 | GATGCCAAAAAA<br>CAAAGGTGAGAA<br>A/CJCCACAAACACA<br>GGTCTAAACTCAG<br>CA | A | C   | SILENT-<br>NONCOD<br>ING | UNCLASSIFIIE<br>D | Human Gene SWISSPROT-<br>ACC:P30968 GONADOTROPIN-<br>RELEASING HORMONE<br>RECEPTOR (GNRH-R) - Homo<br>sapiens (Human), 328 aa.                      | 1.20E-177<br>4 (4q2.2) |
| 388 | cg43924431 | 381  | GTCTTTACAGATG<br>GTTTTCAAAAT'g<br>ap]AGAGTCAGTA<br>AAATAUUCACAIT       | T | gap | SILENT-<br>NONCOD<br>ING | UNCLASSIFIIE<br>D | Human Gene SWISSNEW-<br>ACC:Q16637 SURVIVAL<br>MOTOR NEURON PROTEIN 1 -<br>Homo sapiens (Human), 294 aa.  | 4.20E-166<br>5         |

|     |            |      |  |   |     |                          |                  |  |           |          |
|-----|------------|------|--|---|-----|--------------------------|------------------|--|-----------|----------|
| 389 | cg43936047 | 607  | CGTTGTTCCCTAATG<br>TGGATCTACCAIC/T<br>ICCTGTGTCATC<br>GAGATTCCGGTC     | C | T   | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene TREMBLNEW-<br>ACC:AAD40550 P38P - HOMO<br>SAPIENS (HUMAN), 733 aa.  | 4.30E-164 | 13       |
| 390 | cg43272443 | 1542 | TGGGATTACAGGT<br>GGCACTACACAA<br>/GCCAAAGCTAATT<br>TTGTATTTTAG         | A | G   | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene SWISSNEW-<br>ACC:P13726 TISSUE FACTOR<br>PRECURSOR (TF)<br>(COAGULATION FACTOR III)<br>(THROMBOPLASTIN) (CD142<br>ANTIGEN) - Homo sapiens<br>(Human), 295 aa. | 7.70E-158 | 1 (1p22) |
| 391 | cg43966848 | 2065 | CCTICAGCACCCCT<br>GCAGGGAAAAAC/<br>TAATGAGGCCGG<br>TAGCCGCATCCG        | C | T   | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene SPTREMBL-<br>ACC:Q9Z600 PROTEIN<br>INVOLVED IN SEXUAL<br>DEVELOPMENT, COMPLETE<br>CDS - HOMO SAPIENS<br>(HUMAN), 299 aa.                                      | 4.90E-156 | 2        |
| 392 | cg43964140 | 176  | AAAAAGCTACAGA<br>AAAGAAATCACTT<br>/CTGAAAAAACACA<br>ATGACTCAGAGGC<br>A | T | C   | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene Homologous to<br>TREMBLNEW-ACC:AAC69899<br>SACM21 - MUS MUSCULUS<br>(MOUSE), 721 aa.  | 1.10E-150 | 6        |
| 393 | cg43285114 | 418  | CAGGGACATGGGG<br>GCACCCCGGGGG<br>/gap/CTTGGGGC<br>TCACAGGACAATG<br>G   | G | gap | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene Homologous to<br>TREMBLNEW-ACC:AAD23440<br>LR8 - HOMO SAPIENS<br>(HUMAN), 270 aa.   | 1.90E-138 | 7        |

|     |            |     |   |   |     |  |                          |                  |   |           |    |
|-----|------------|-----|---|---|-----|--|--------------------------|------------------|---|-----------|----|
| 394 | cgt3948566 | 370 | GCAGGGCAAGCAC<br>CCTGGGACCCCAIG<br>[gap]GGCAAGAGGA<br>CCCCTGCCCTCCAG<br>T | G | gap |  | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene Homologous to<br>SWISSNEW-ACC:P18382 CD81<br>ANTIGEN (26 KD CELL<br>SURFACE PROTEIN TAPA-1)-<br>Homo sapiens (Human), 236 aa.  | 3.30E-125 | 11 |
| 395 | cgt4003626 | 649 | TAAACAGCTCAGT<br>TCAGGGACTGGT[A<br>G]TACAAGCTGGC<br>CACCCATCTCAGC<br>C    | A | G   |  | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene Homologous to<br>SPTREMBL-ACC:Q15025 mRNA<br>(HA1652) FOR ORF, PARTIAL<br>CDS - HOMO SAPIENS<br>(HUMAN), 296 aa (fragment).  | 2.70E-123 |    |
| 396 | cgt3917206 | 259 | TTACAGGACATCA<br>CCTGCCATCTTAIT/<br>AIGGTTAATATT<br>ACAAATGCCTAGT         | T | A   |  | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene Homologous to<br>SWISSPROT-ACC:P22061<br>PROTEIN-L-ISOASPARTATE(D-<br>ASPARTATE) O-<br>METHYLTRANSFERASE (EC<br>2.1.1.77) (PROTEIN-BETA-<br>ASPARTATE<br>METHYLTRANSFERASE)<br>(PIMT) (PROTEIN L-<br>ISOASPARTYL/D-ASPARTYL<br>METHYLTRANSFERASE) (L-<br>ISOASPARTYL PROTEIN<br>CARBOXYL<br>METHYLTRANSFERASE)-<br>Homo sapiens (Human), 226 aa. | 6.90E-118 | 6  |

|     |            |      |  |   |   |                          |                  |   |           |                     |
|-----|------------|------|--|---|---|--------------------------|------------------|---|-----------|---------------------|
| 397 | cg43289666 | 215  | GGCCGATTTCCA<br>CAATTAAATC[CT<br>[CAGTTAACCTGGT<br>ATCCAGTCCAG     | C | T | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene Homologous to<br>SPTREMBL-ACC:Q00539<br>CANCER ASSOCIATED<br>SURFACE ANTIGEN - HOMO<br>SAPIENS (HUMAN), 213 aa.  | 2.50E-111 | 8                   |
| 398 | cg43986282 | 840  | GTTCCACCTCCCC<br>AGACAGGCCATT[C<br>T]CGAGTGGGAGGC<br>GGGAGCACGTACC | C | T | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene Homologous to<br>SPTREMBL-ACC:P97314<br>DOUBLE LIM PROTEIN-1 -<br>MUS MUSCULUS (MOUSE), 193<br>aa.   | 2.90E-110 | 12                  |
| 399 | cg43986282 | 841  | TTTCCACCTCCCC<br>GACAGGCCATT[C<br>T]GAGTGGGAGGC<br>GGGAGCACGTACC   | C | T | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene Homologous to<br>SPTREMBL-ACC:P97314<br>DOUBLE LIM PROTEIN-1 -<br>MUS MUSCULUS (MOUSE), 193<br>aa.   | 2.90E-110 | 12                  |
| 400 | cg43297716 | 1030 | CTAAACCAAATG<br>GGGGCTGTCGGC[A<br>T]GACCCCGAGGG<br>TGCTGGCCAGTC    | A | T | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene Homologous to<br>SWISSPROT-ACC:P15018<br>LEUKEMIA INHIBITORY<br>FACTOR PRECURSOR (LIF)<br>(DIFFERENTIATION-<br>STIMULATING FACTOR) (D<br>FACTOR) (MELANOMA-<br>DERIVED LPL INHIBITOR)<br>(MLPL) - Homo sapiens (Human),<br>202 aa. | 1.20E-106 | 22<br>(22q12.1<br>) |

|     |            |      |   |          |  |                           |                   |   |           |         |
|-----|------------|------|---|----------|--|---------------------------|-------------------|---|-----------|---------|
| 401 | cg43980312 | 2160 | TITATCATTAAAG<br>TGCCAGAATGGC/<br>TCTTAATGAAA<br>ACAAAAACAAAG           | C<br>T   |  | SILENT-<br>NONCOD-<br>ING | UNCLASSIFI-<br>ED | Human Gene Homologous to<br>SWISSPROT-ACC:P34741<br>SYNDECAN-2 PRECURSOR<br>(FIBROGL YCAN) (HEPARAN<br>SULFATE PROTEOGLYCAN<br>CORE PROTEIN) (HSPG)<br>(SYND2) - Homo sapiens<br>(Human), 201 aa.                           | 7.90E-101 | 8 (8q2) |
| 402 | cg43939240 | 624  | GGAGGGTTGGACT<br>CACTGACCAATGIC<br>/T]AGCCCCGCCA<br>GGCCCATGCAAAG<br>G  | C<br>T   |  | SILENT-<br>NONCOD-<br>ING | UNCLASSIFI-<br>ED | Human Gene Similar to<br>SPTREMBL-ACC:Q43399<br>HD54+DNS2 ISOFORM - HOMO<br>SAPIENS (HUMAN), 206 aa.  | 1.00E-100 |         |
| 403 | cg43941552 | 881  | GCCACCTGCCCG<br>GCTGGAGGGAG[C<br>/gap]GCTGGCGCTG<br>ACCAAGGGCTGGG<br>GC | C<br>gap |  | SILENT-<br>NONCOD-<br>ING | UNCLASSIFI-<br>ED | Human Gene Similar to<br>SWISSNEW-ACC:P11686<br>PULMONARY SURFACTANT-<br>ASSOCIATED PROTEIN C<br>PRECURSOR (SP-C) (SP5)<br>(PULMONARY SURFACTANT-<br>ASSOCIATED PROTEOLIPID<br>SPL(VAL)) - Homo sapiens<br>(Human), 197 aa. | 1.60E-100 |         |
| 404 | cg43941552 | 1124 | GCTCTGCCACAC<br>CGCAGGGACAA[A/<br>G]CCCTGGAGAAAT<br>GGGAGCNGGGA         | A<br>G   |  | SILENT-<br>NONCOD-<br>ING | UNCLASSIFI-<br>ED | Human Gene Similar to<br>SWISSNEW-ACC:P11686<br>PULMONARY SURFACTANT-<br>ASSOCIATED PROTEIN C<br>PRECURSOR (SP-C) (SP5)<br>(PULMONARY SURFACTANT-<br>ASSOCIATED PROTEOLIPID<br>SPL(VAL)) - Homo sapiens<br>(Human), 197 aa. | 1.60E-100 |         |

|     |            |     |   |   |   |                  |              |   |           |    |
|-----|------------|-----|---|---|---|------------------|--------------|---|-----------|----|
| 405 | cg42917153 | 914 | CATTCTCTTGTACATAATACATTCTAACCTCCCTGCCCTCTCTCCTTCTA                  | C | T | SILENT-NONCODING | UNCLASSIFIED | Human Gene Similar to SWISSPROT-ACC:P45973 HETEROCHROMATIN PROTEIN 1 HOMOLOG ALPHA (HP1 ALPHA) (ANTIGEN P25) - Homo sapiens (Human), 191 aa.                            | 2.10E-100 | 12 |
| 406 | cg43927693 | 878 | CAGGGGTAGGCAGAGCTCAGAGGTGTTGCCCAACCTGA<br>A                         | G | T | SILENT-NONCODING | UNCLASSIFIED | Human Gene Similar to SWISSPROT-ACC:P30536 PERIPHERAL-TYPE BENZODIAZEPINE RECEPTOR (PBR) (PKBS) (MITOCHONDRIAL BENZODIAZEPINE RECEPTOR) - Homo sapiens (Human), 169 aa. | 5.30E-95  | 22 |
| 407 | cg43951338 | 507 | CAGAAAGCAGCAA<br>ATTAGCTTTTC/ AAGGACCGAATTCCGCTCCGGAGCT             | C | A | SILENT-NONCODING | UNCLASSIFIED | Human Gene Similar to SWISSPROT-ACC:P36405 ADP-RIBOSYLATION FACTOR-LIKE PROTEIN 3 - Homo sapiens (Human), 182 aa.   | 3.40E-93  | 10 |
| 408 | cg43951338 | 511 | AAGCAGCAAATTAGTGTTCAGG/C<br>CJCCGAATTGGCT<br>CCCGCAGCTCCTG          | A | C | SILENT-NONCODING | UNCLASSIFIED | Human Gene Similar to SWISSPROT-ACC:P36405 ADP-RIBOSYLATION FACTOR-LIKE PROTEIN 3 - Homo sapiens (Human), 182 aa.   | 3.40E-93  | 10 |
| 409 | cg43951338 | 547 | CTCCCGCAGCTCCT<br>GCATCTCCATTTC/T<br>IGCTCTAGATTCTAT<br>TCTCTCTTGCA | C | T | SILENT-NONCODING | UNCLASSIFIED | Human Gene Similar to SWISSPROT-ACC:P36405 ADP-RIBOSYLATION FACTOR-LIKE PROTEIN 3 - Homo sapiens (Human), 182 aa.   | 3.40E-93  | 10 |

|     |            |      |  |     |   |  |                          |                  |   |          |
|-----|------------|------|--|-----|---|--|--------------------------|------------------|---|----------|
| 410 | cg25236776 | 1234 | CCGGCCCAAGCCCC<br>ACGCCTACTGTAGA<br>ap/TCCCCCGGCGTC<br>GCCCAACGGGCGC<br>GC | gap | T |  | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene Similar to<br>SWISSNEW-ACC;P01185<br>VASOPRESSIN-NEUROPHYSIN<br>2-COPEPTIN PRECURSOR<br>[CONTAINS: ARG-<br>VASOPRESSIN; NEUROPHYSIN<br>2 (NEUROPHYSIN-II);<br>COPEPTIN] - Homo sapiens<br>(Human), 164 aa. | 7.20E-91 |
| 411 | cg25236776 | 1240 | CCAGCCCGAACGCC<br>TACIGAGCCCCGIC<br>/T[GCTGCCCCACC<br>GGCGCGCTCTCG         | C   | T |  | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene Similar to<br>SWISSNEW-ACC;P01185<br>VASOPRESSIN-NEUROPHYSIN<br>2-COPEPTIN PRECURSOR<br>[CONTAINS: ARG-<br>VASOPRESSIN; NEUROPHYSIN<br>2 (NEUROPHYSIN-II);<br>COPEPTIN] - Homo sapiens<br>(Human), 164 aa. | 7.20E-91 |
| 412 | cg25236776 | 1242 | AGCCGACGCC<br>CTGAGCCCCCGCIC<br>/T[TGCCCCCACCGG<br>CGGCCTCTCGCG            | C   | T |  | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene Similar to<br>SWISSNEW-ACC;P01185<br>VASOPRESSIN-NEUROPHYSIN<br>2-COPEPTIN PRECURSOR<br>[CONTAINS: ARG-<br>VASOPRESSIN; NEUROPHYSIN<br>2 (NEUROPHYSIN-II);<br>COPEPTIN] - Homo sapiens<br>(Human), 164 aa. | 7.20E-91 |
| 413 | cg25236776 | 1246 | CGACGCCTACTGA<br>GCCCGGGCTCGIC<br>/T]CCCACGGCGC<br>GCTCTCGCGCCCG           | C   | T |  | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>ED | Human Gene Similar to<br>SWISSNEW-ACC;P01185<br>VASOPRESSIN-NEUROPHYSIN<br>2-COPEPTIN PRECURSOR<br>[CONTAINS: ARG-<br>VASOPRESSIN; NEUROPHYSIN<br>2 (NEUROPHYSIN-II);<br>COPEPTIN] - Homo sapiens<br>(Human), 164 aa. | 7.20E-91 |

|     |            |      |  |     |     |  |                          |                 |   |          |                     |
|-----|------------|------|--|-----|-----|--|--------------------------|-----------------|---|----------|---------------------|
| 414 | cg43968406 | 1362 | GCTACGTTTACTCA<br>CAGCCAGGGAA[ga<br>p[A]CTGACATTTAA<br>ATAACTAACAAAC<br>A  | gap | A   |  | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>E | Human Gene Similar to<br>REMBL-ACC:E47283<br>DNA FOR ORF1 AND ORF2<br>FROM CHROMOSOME X -<br>HOMO SAPIENS (HUMAN), 157<br>aa. | 5.00E-83 | X<br>(Xp11.4)       |
| 415 | cg42748886 | 104  | CGCCTCTCATCCA<br>AGCCACCTCCCGIC<br>TTCAGAGGGTGT<br>CATGGGCTTCCAA<br>A      | C   | T   |  | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>E | Human Gene Similar to<br>SWISSNEW-ACC:P01258<br>CALCITONIN PRECURSOR •<br>Homo sapiens (Human), 141 aa.                       | 2.00E-70 | 11<br>(11p15.2<br>) |
| 416 | cg43969533 | 356  | CTCTGCACAAAGGG<br>AAGCCTATCCTAT/<br>gap]TTTTTTTCT<br>TTGCAAAAACAGA         | T   | gap |  | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>E | Human Gene Similar to<br>REMBLNEW-ACC:AAD39844<br>HSPC028 - HOMO SAPIENS<br>(HUMAN), 419 aa.                                  | 1.60E-67 | 7                   |
| 417 | cg43976681 | 1119 | AATGCCCTCAGATC<br>AGTGACCCAAAGG]<br>A/gap]ACCTTCAG<br>AATGGATGAAATA<br>GAC | A   | gap |  | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>E | Human Gene Similar to<br>REMBLNEW-ACC:AAD29427<br>MYOMEGLIN - RATTUS<br>NORVEGICUS (RAT), 2324 aa.                            | 4.30E-66 | 11                  |
| 418 | cg43976681 | 1120 | ATGCCTCAGATCA<br>GTGACCCAAAGGA[<br>A/gap]CCTTCAGA<br>ATGGATGAAATA<br>ACC   | A   | gap |  | SILENT-<br>NONCOD<br>ING | UNCLASSIFI<br>E | Human Gene Similar to<br>REMBLNEW-ACC:AAD29427<br>MYOMEGLIN - RATTUS<br>NORVEGICUS (RAT), 2324 aa.                            | 4.30E-66 | 11                  |

|     |            |     |   |        |  |                          |              |   |          |          |
|-----|------------|-----|---|--------|--|--------------------------|--------------|---|----------|----------|
| 419 | cg43984044 | 714 | CCAAGCGGAAGGC<br>CATTTCCTGC/C/<br>TCTTCCTCAGTIG<br>TCCGGGGGGGG        | C<br>T |  | SILENT-<br>NONCOD<br>ING | UNCLASSIFIIE | Human Gene Similar to<br>SPTREMBL-ACC:Q00455 TTf-1<br>INTERACTING PEPTIDE 20 -<br>HOMO SAPIENS (HUMAN), 385<br>aa (fragment).   | 7.30E-66 | 19       |
| 420 | cg43933283 | 398 | CTAATTTGTCGA<br>ATTCAGGATT[G/<br>A]AGGAAAAGTT<br>GCTCCCTTCAGCC        | G<br>A |  | SILENT-<br>NONCOD<br>ING | UNCLASSIFIIE | Human Gene Similar to<br>SWISSPROT-ACC:P05062<br>FRUCTOSE-BISPHOSPHATE<br>ALDOLASE B (EC 4.1.2.13)<br>(LIVER-TYPE ALDOLASE) -<br>Homo sapiens (Human), 363 aa.          | 6.60E-65 | 9 (9q22) |
| 421 | cg42381630 | 577 | AAAGCAATCACAG<br>TGTTAAAGAAGIG<br>[A]CAGCTGAAAT<br>GATGCCAGCTGCT<br>C | G<br>A |  | SILENT-<br>NONCOD<br>ING | UNCLASSIFIIE | Human Gene Similar to<br>SPTREMBL-ACC:Q76087<br>GAGE-8 - HOMO SAPIENS<br>(HUMAN), 117 aa.   | 5.90E-64 |          |
| 422 | cg41664708 | 423 | CCAGCCAGCTCAT<br>TTCACTTACAC[G/<br>C]CTCATGACTGA<br>GTTTATACTCACC     | G<br>C |  | SILENT-<br>NONCOD<br>ING | UNCLASSIFIIE | Human Gene Similar to<br>SWISSNEW-ACC:P47992<br>LYMPHOTACTIN PRECURSOR<br>(CYTOKINE SCM-1) (ATAC)<br>(LYMPHOTAXIN) (SCM-1-<br>ALPHA) - Homo sapiens (Human),<br>114 aa. | 2.00E-54 | 1        |

|     |            |      |  |   |   |     |              |                  |                        |  |   |                     |
|-----|------------|------|--|---|---|-----|--------------|------------------|------------------------|--|---|---------------------|
| 423 | cg43277632 | 3906 | AGCCTTCGGCAG<br>AAAAAGATGCA<br>CTCCCCAGACCT<br>TCCTGTGCTGATT           | C | T | Ala | Val<br>(652) | CONSER<br>VATIVE | ATPase _associat<br>ed | Human Gene SWISSPROT-<br>ID:P5670 COPPER-<br>TRANSPORTING ATPASE 2 (EC<br>3.6.1.36) (COPPER PUMP 2)<br>(WILSON DISEASE-<br>ASSOCIATED PROTEIN)-<br>HOMO SAPIENS (HUMAN)-<br>1465 aa. | 0 | 13<br>(13q14.3<br>) |
| 424 | cg40310734 | 1138 | TACCAAGAGGCTGC<br>ATCGGCTGCGCGC<br>/GIAAGAGCAGATGG<br>CGTCGTATTTGGG    | C | G | Ala | Gly<br>(653) | CONSER<br>VATIVE | catenin                | Human Gene SWISSPROT-<br>ID:PO8514 PLATELET<br>MEMBRANE GLYCOPROTEIN<br>IIb PRECURSOR (GP1IB)<br>(INTEGRIN ALPHA- IIb) (CD41)<br>-HOMO SAPIENS (HUMAN),<br>1039 aa.                  | 0 | 17<br>(17q21.3<br>) |
| 425 | cg40310734 | 1238 | TGGTGGGGCTCC<br>ACTGTATATGGAIG<br>/CJAGCCGGCAGA<br>CCGAAAACGGCC<br>G   | G | C | Glu | Asp<br>(654) | CONSER<br>VATIVE | catenin                | Human Gene SWISSPROT-<br>ID:PO8514 PLATELET<br>MEMBRANE GLYCOPROTEIN<br>IIb PRECURSOR (GP1IB)<br>(INTEGRIN ALPHA- IIb) (CD41)<br>-HOMO SAPIENS (HUMAN),<br>1039 aa.                  | 0 | 17<br>(17q21.3<br>) |
| 426 | cg40310734 | 1893 | CTCTCAAAGGCCA<br>GGCACCAACCTG[A<br>/G]ACCTGGATCTG<br>GGGGAAAGCACA<br>G | A | G | Asn | Asp<br>(655) | CONSER<br>VATIVE | catenin                | Human Gene SWISSPROT-<br>ID:PO8514 PLATELET<br>MEMBRANE GLYCOPROTEIN<br>IIb PRECURSOR (GP1IB)<br>(INTEGRIN ALPHA- IIb) (CD41)<br>-HOMO SAPIENS (HUMAN),<br>1039 aa.                  | 0 | 17<br>(17q21.3<br>) |

|     |            |      |   |   |              |                     |                  |              |   |           |                     |
|-----|------------|------|---|---|--------------|---------------------|------------------|--------------|---|-----------|---------------------|
| 427 | cg43982507 | 1883 | GGTTACAAGTGTG<br>AATGTAAGTCGTG<br>/CCTATCAAATGG<br>ATCTTGCTACTGGC       | G | C            | Gly<br>Ala<br>(656) | CONSER<br>VATIVE | eph          | Human Gene SWISSPROT-<br>ID:P08155 VERY LOW-<br>DENSITY LIPOPROTEIN<br>RECEPTOR PRECURSOR<br>(VLDL RECEPTOR) - HOMO<br>SAPIENS (HUMAN), 873 aa.   | 0         | 9 (9p24)            |
| 428 | cg41554010 | 949  | GCGAGGGACGTGC<br>GTGGCAACCTGA[G<br>/AAGGCCAACACCG<br>AGGGGGCTGAGAA<br>G | A | Arg<br>(657) | Lys<br>(657)        | CONSER<br>VATIVE | eph          | Human Gene SWISSNEW-<br>ID:P06727 APOLIPOPROTEIN A-<br>IV PRECURSOR (APO-AIV) -<br>HOMO SAPIENS (HUMAN), 396<br>aa [pcds;SWISSPROT ID:P06727]<br>APOLIPOPROTEIN A-IV<br>PRECURSOR (APO-AIV) -<br>HOMO SAPIENS (HUMAN), 396<br>aa. | 1.80E-203 | 11<br>(11q23)       |
| 429 | cg43299024 | 1036 | TACGGAGGTGCCCT<br>TGGAGACCCGGCA<br>/GTGTCCACAGCC<br>GGGCACCGTCCCC<br>A  | A | G            | His<br>(658)        | CONSER<br>VATIVE | glucoamylase | Human Gene TREMBL NEW-<br>ID:G2826521 MALTASE-<br>GLUCOAMYLASE (EC 3.2.1.20)<br>- HOMO SAPIENS (HUMAN),<br>1857 aa.   | 7.40E-199 | 17<br>(17q25.2<br>) |
| 430 | cg43299024 | 1108 | GAGGAGCCCTTCG<br>GGGTGATCGTCGA<br>/GICCGGGAGCTGG<br>ACGGCCGCGTGCT<br>G  | A | G            | His<br>(659)        | CONSER<br>VATIVE | glucoamylase | Human Gene TREMBL NEW-<br>ID:G2826521 MALTASE-<br>GLUCOAMYLASE (EC 3.2.1.20)<br>- HOMO SAPIENS (HUMAN),<br>1857 aa.   | 7.40E-199 | 17<br>(17q25.2<br>) |

|     |            |       |   |   |   |     |              |                  |              |  |          |                     |
|-----|------------|-------|---|---|---|-----|--------------|------------------|--------------|--|----------|---------------------|
| 431 | cg43285373 | 12840 | GAATGACTGGGAA<br>AAGGAACCTAAAC<br>A/CCTCGAGTCGCC<br>TGGATGAATGGAG<br>A  | A | C | Ile | Leu<br>(660) | CONSER<br>VATIVE | glycoprotein | Human Gene SWISSPROT-<br>ID:P98164 LOW-DENSITY<br>LIPOPROTEIN RECEPTOR-<br>RELATED PROTEIN 2<br>(MEGALIN) (GLYCOPROTEIN<br>330) - HOMO SAPIENS<br>(HUMAN), 1751 aa (fragment).         | 0        | 2                   |
| 432 | cg46834323 | 1004  | AGTTATTCTTAGAG<br>GATACAGAAATCIA<br>(G)TCGAAGTCCC<br>GAGAAACTAGGGA<br>G | A | G | His | Arg<br>(661) | CONSER<br>VATIVE | glycoprotein | Human Gene Similar to<br>SWISSPROT-ID:P38159<br>HETEROGENEOUS NUCLEAR<br>RIBONUCLEOPROTEIN G<br>(HNRNP G) (GLYCOPROTEIN<br>P43) - HOMO SAPIENS<br>(HUMAN), 437 aa.                     | 6.40E-91 |                     |
| 433 | cg41568631 | 2101  | GGACCAGGGGGCC<br>ATGCTGCTCAATG/<br>ATTCCTCAGGCCACG<br>TCAAGGAGAGCGG     | G | A | Val | Ile<br>(662) | CONSER<br>VATIVE | glycoprotein | Human Gene Similar to<br>SWISSPROT-ID:P16452<br>ERYTHROCYTE MEMBRANE<br>PROTEIN BAND 4.2 (P4.2)<br>(PALLIDIN) - HOMO SAPIENS<br>(HUMAN), 690 aa.                                       | 9.90E-70 | 14<br>(14q11.2<br>) |
| 434 | cg42359655 | 666   | TGCTTTCAAGGGCG<br>GAAAACCTCTTA/<br>GTTTGCTCTGCAG<br>CTGAAGATAATCCC      | A | G | Ile | Val<br>(663) | CONSER<br>VATIVE | hydrolase    | Human Gene SWISSPROT-<br>ID:P09848 LACTASE-<br>PHLORIZIN HYDROLASE<br>PRECURSOR (EC 3.2.1.108) (EC<br>3.2.1.62) (LACTASE-<br>GLYCOSYLCERAMIDASE)-<br>HOMO SAPIENS (HUMAN),<br>1927 aa. | 0        | 2 (241)             |

|     |            |      |  |                        |              |                  |                  |   |  |                     |               |
|-----|------------|------|--|------------------------|--------------|------------------|------------------|---|--|---------------------|---------------|
| 435 | cg43998672 | 1331 | GTGTGGCCCTTGG<br>TGAACCTAGCAIC<br>AIGCGGGCTAAATGT<br>CTCCTGGTTGGTC | C<br>A<br>A            | Val<br>(664) | Leu<br>(664)     | CONSER<br>VATIVE | hydroxysteroid  | Human Gene SPTREMBL-<br>ID:Q13194 11-BETA-<br>HYDROXYSTEROID<br>DEHYDROGENASE TYPE 2 -<br>HOMO SAPIENS (HUMAN), 405<br>aa. | 2.00E-220           | 16<br>(16q22) |
| 436 | cg43969028 | 1133 | GGAGATGGGTCA<br>TTCCTAGTGATTAA<br>TTTTTCAGATAGT<br>GGGAGGAAGCAAC   | A<br>T<br>Tyr<br>(665) | Phe<br>(665) | CONSER<br>VATIVE | immunoglob       | Human Gene Homologous to<br>SPTREMBL-ID:P91456 SIMILAR<br>TO THE IMMUNOGLOBULIN<br>SUPERFAMILY -<br>CAENORHABDITIS ELEGANS,<br>1173 aa. | 2.20E-149  | 18<br>(18q21.3<br>) |               |

|     |            |      |   |   |   |              |              |                  |             |   |           |    |
|-----|------------|------|---|---|---|--------------|--------------|------------------|-------------|---|-----------|----|
| 437 | cgt3933479 | 133  | AAGGAGAAGAGAA<br>AGCTGTTATCCJA/<br>GTTCCATGGTGA<br>AGGTACATAAAT | A | G | His<br>(666) | Arg<br>(666) | CONSER<br>VATIVE | interleukin | Human Gene SWISSNEW-<br>ID:P29466 INTERLEUKIN-1<br>BETA CONVERTASE<br>PRECURSOR (IL-1 BCF) (EC<br>3.4.22.36) (IL-1 BETA<br>CONVERTING ENZYME) (ICE)<br>(INTERLEUKIN-1 BETA<br>CONVERTING ENZYME) (P45)<br>(CASPASE-1) (CASP-1) - HOMO<br>SAPIENS (HUMAN), 404<br>aa.lpcis:SWISSPROT-ID:P29466<br>INTERLEUKIN-1 BETA<br>CONVERTASE PRECURSOR<br>(IL-1 BCF) (EC 3.4.22.36) (IL-1<br>BETA CONVERTING ENZYME)<br>(ICE) (INTERLEUKIN-1 BETA<br>CONVERTING ENZYME) (P45)<br>(CASPASE-1) (CASP-1) - HOMO<br>SAPIENS (HUMAN), 404 aa. | 2.50E-206 |    |
| 438 | cgt3942537 | 1163 | GCCACTGTCTTC<br>CAAACCCCTCA[<br>A]GCCCTTGCTTG<br>TTGTTCTCGTCTA  | C | A | Val<br>(667) | Leu<br>(667) | CONSER<br>VATIVE | kinesin     | Human Gene SWISSNEW-<br>ID:P33176 KINESIN HEAVY<br>CHAIN (UBIQUITOUS KINESIN<br>HEAVY CHAIN) (UKHC) -<br>HOMO SAPIENS (HUMAN), 963<br>aa.lpcis:SWISSPROT-ID:P33176<br>KINESIN HEAVY CHAIN<br>(UBIQUITOUS KINESIN<br>HEAVY CHAIN) (UKHC) -<br>HOMO SAPIENS (HUMAN), 963<br>aa.   | 0         | 10 |

|     |            |      |  |   |   |              |                  |             |  |           |               |
|-----|------------|------|--|---|---|--------------|------------------|-------------|--|-----------|---------------|
| 439 | cg38337333 | 1035 | TTCCAATGCTGA<br>GCCAGAGCGTTG<br>/ATCTCCTGCCAT<br>GAGCACACAGTC        | G | A | Val<br>(668) | CONSER<br>VATIVE | MHC         | Human Gene Homologous to<br>SPTREMBL-ID:Q95368 HLA<br>CLASS I INHIBITORY NK<br>RECEPTOR - HOMO SAPIENS<br>(HUMAN), 455 aa.   | 1.80E-113 | 19            |
| 440 | cg38337333 | 271  | CTGGAAACAGTTTC<br>CTCATTAGCCCTTG/<br>CTTGACCCAGCAC<br>ACGCAGGGACCTA  | G | C | Val<br>(669) | CONSER<br>VATIVE | MHC         | Human Gene Homologous to<br>SPTREMBL-ID:Q95368 HLA<br>CLASS I INHIBITORY NK<br>RECEPTOR - HOMO SAPIENS<br>(HUMAN), 455 aa.   | 1.80E-113 | 19            |
| 441 | cg38337333 | 823  | TCAATGGCTGGTGT<br>CCA AAAAAAA[A/<br>G]ATGGCTGGTGTAA<br>TGAACCAAGAGCC | A | G | Asn<br>(670) | CONSER<br>VATIVE | MHC         | Human Gene Homologous to<br>SPTREMBL-ID:Q95368 HLA<br>CLASS I INHIBITORY NK<br>RECEPTOR - HOMO SAPIENS<br>(HUMAN), 455 aa.   | 1.80E-113 | 19            |
| 442 | cg30421838 | 3434 | GGATGCTGTTGCTC<br>TCCCACAGCCAG/<br>TTGGGGCGTTCAA<br>ATGAAAAGCCAAGC   | G | T | Val<br>(671) | CONSER<br>VATIVE | nucl_recept | Human Gene SWISSNEW-<br>ID:P06401 PROGESTERONE<br>RECEPTOR (PR) - HOMO<br>SAPIENS (HUMAN), 933<br>aa.[pcl:SWISSPROT-ID:P06401<br>PROGESTERONE RECEPTOR<br>(PR) - HOMO SAPIENS<br>(HUMAN), 933 aa.] | 0         | II<br>(11q22) |

|     |            |      |   |   |   |     |              |                  |             |   |           |               |
|-----|------------|------|---|---|---|-----|--------------|------------------|-------------|---|-----------|---------------|
| 443 | cg43064060 | 1019 | GCCAATGGCATCC<br>AGAACAAAGGAGGI<br>CTTGGAGGCCGGC<br>ATCTTTCACTGCTG<br>C | C | T | Ala | Val<br>(672) | CONSER<br>VATIVE | nucl_recept | Human Gene SWISSPROT-<br>ID:Q07869 PEROXISOME<br>PROLIFERATOR ACTIVATED<br>RECEPTOR ALPHA (PPAR-<br>ALPHA) - HOMO SAPIENS<br>(HUMAN), 468<br>aa. Ipcls:SPTREMBL-ID:Q16241 | 4.10E-254 | 22            |
| 444 | cg43991813 | 1860 | TCTCGACTAACAG<br>CATTCCAAGAT/<br>CIGGAGCGAATATT<br>GTCCACGGTTGAG        | T | C | Ile | Val<br>(673) | CONSER<br>VATIVE | nuclease    | Human Gene SWISSPROT-<br>ID:P40692 MUTL PROTEIN<br>HOMOLOG 1 (DNA MISMATCH<br>REPAIR PROTEIN MLH1)-<br>HOMO SAPIENS (HUMAN), 756<br>aa.                                   | 0         | 3<br>(3p21.3) |
| 445 | cg42904626 | 194  | GAGTCCTTGACG<br>ATACAGCTAATTIC/<br>GJAGAAATCATTTG<br>TGGACGAATATGA      | C | G | Gln | Glu<br>(674) | CONSER<br>VATIVE | oncogene    | Human Gene Similar to<br>SWISSPROT-ID:PO1118<br>TRANSFORMING PROTEIN<br>P21/K-RAS 2B - HOMO<br>SAPIENS (HUMAN), 188 aa.   | 1.10E-97  | 12            |
| 446 | cg42904626 | 548  | AAGAAAGTTATGGA<br>ATTCCTTTATTG/<br>CIAAACATCAGCAA<br>AGACAAGACAGGG      | G | C | Glu | Gln<br>(675) | CONSER<br>VATIVE | oncogene    | Human Gene Similar to<br>SWISSPROT-ID:PO1118<br>TRANSFORMING PROTEIN<br>P21/K-RAS 2B - HOMO<br>SAPIENS (HUMAN), 188 aa.   | 1.10E-97  | 12            |

|     |            |      |  |   |   |     |              |                  |             |  |          |               |
|-----|------------|------|--|---|---|-----|--------------|------------------|-------------|--|----------|---------------|
| 447 | cg42460457 | 2845 | GCCGCCCTCAGCCA<br>GCAAGCAGGGGI<br>CTTAGGCCAGTCC<br>TAGCCACACAGA<br>G | C | T | Ala | Val<br>(676) | CONSER<br>VATIVE | phosphatase | Human Gene SWISSPROT-<br>ID:P23470 PROTEIN-TYROSINE<br>PHOSPHATASE GAMMA<br>PRECURSOR (EC 3.1.3.48) (R-<br>PTP-GAMMA) - HOMO<br>SAPIENS (HUMAN), 1445 aa.  | 0        | 3<br>(3p14.2) |
| 448 | cg43272594 | 582  | GGGATGTACTGCA<br>TGGTGTTCCTGGGT/<br>CJGCCTGTATGTGCA<br>GGCACGACTCTGT | T | C | Val | Ala<br>(677) | CONSER<br>VATIVE | phosphatase | Human Gene Similar to<br>SPTREMBL-ID:Q61469<br>PHOSPHATIC ACID<br>PHOSPHATASE - MUS<br>MUSCUS (MOUSE), 283 aa.   | 1,40E-79 | 19            |
| 449 | cg43958858 | 807  | TCAGGTGGTGGGA<br>ACCTACCGTTGCIC/<br>GJTCCCTGAAAGA<br>AGGGAGGCTACAC   | C | G | Leu | Val<br>(678) | CONSER<br>VATIVE | polymerase  | Human Gene SWISSNEW-<br>ID:P25205 DNA REPLICATION<br>LICENSING FACTOR MCM3<br>(DNA POLYMERASE ALPHA<br>HOLOENZYME-ASSOCIATED<br>PROTEIN P1) (RLF BETA<br>SUBUNIT) (P102 PROTEIN) -<br>HOMO SAPIENS (HUMAN), 808<br>aa. Ipcis:SWISSPROT-ID:P25205<br>DNA REPLICATION<br>LICENSING FACTOR MCM3<br>(DNA POLYMERASE ALPHA<br>HOLOENZYME-ASSOCIATED<br>PROTEIN P1) (RLF BETA<br>SUBUNIT) (P102 PROTEIN) -<br>HOMO SAPIENS (HUMAN), 808<br>aa. | 0        | 6<br>(6p12)   |

|     |            |      |  |   |   |              |              |                  |          |   |                       |                 |
|-----|------------|------|--|---|---|--------------|--------------|------------------|----------|---|-----------------------|-----------------|
| 450 | cg43916732 | 540  | GTACAGGGGGGG<br>GCCACCTCGGGC[A<br>T]CTGAGCACCAA<br>TTTGGGGGGC<br>G       | A | T | Thr<br>(679) | Ser<br>(679) | CONSER<br>VATIVE | protease | Human Gene SPTREM BL-<br>ID:Q15113 PROCOLLAGEN C-<br>PROTEINASE ENHANCER<br>PROTEIN PRECURSOR - HOMO<br>SAPIENS (HUMAN), 449 aa.                                      | 1.20E-247<br>(7q21.3) | 7               |
| 451 | cg42894809 | 2745 | GGATGGTGGAGAG<br>TGATCATCTGTCIA/<br>GJATCAGACGACA<br>ACAGCCAAACCGTT<br>A | A | G | Asn<br>(680) | Asp<br>(680) | CONSER<br>VATIVE | struct   | Human Gene SWISSPROT-<br>ID:P54296 M-PROTEIN (165 KD<br>TITIN-ASSOCIATED PROTEIN)<br>(165 KD CONNECTIN-<br>ASSOCIATED PROTEIN) -<br>HOMO SAPIENS (HUMAN),<br>1465 aa. | 0                     | 8               |
| 452 | cg40388639 | 2337 | GATTCTCCAGAG<br>CTGGTGTGGAAIG<br>[C]TTCCCATCAGGC<br>ACCCAAGTTGA          | G | C | Val<br>(681) | Leu<br>(681) | CONSER<br>VATIVE | synthase | Human Gene SWISSPROT-<br>ID:P29475 NITRIC-OXIDE<br>SYNTHASE, BRAIN (EC<br>1.14.13.39) (NOS, TYPE I)<br>(NEURONAL NOS) (NNOS) -<br>HOMO SAPIENS (HUMAN),<br>1434 aa.   | 0                     | 12<br>(12q24.2) |
| 453 | cg40388639 | 2380 | AAGTTGAGTGGT<br>TCAAGGACCTGGIG<br>[C]GCTGAAGTGGT<br>ACGGCCCTCCCCGC<br>C  | G | C | Gly<br>(682) | Ala<br>(682) | CONSER<br>VATIVE | synthase | Human Gene SWISSPROT-<br>ID:P29475 NITRIC-OXIDE<br>SYNTHASE, BRAIN (EC<br>1.14.13.39) (NOS, TYPE I)<br>(NEURONAL NOS) (NNOS) -<br>HOMO SAPIENS (HUMAN),<br>1434 aa.   | 0                     | 12<br>(12q24.2) |

|     |            |      |   |                  |                     |                  |          |   |          |    |
|-----|------------|------|---|------------------|---------------------|------------------|----------|---|----------|----|
| 454 | cg43124627 | 1524 | AATTCTATATCAC<br>TGGGACAGAGIC/<br>GJATATAGGATAA<br>AGATGGTATTTC     | C<br>G<br>C<br>G | Ala<br>Gly<br>(683) | CONSER<br>VATIVE | synthase | Human Gene Similar to<br>SWISSNEW-ID:P39062<br>ACETYL-COENZYME A<br>SYNTHETASE (EC 6.2.1.1)<br>(ACETATE-COA LIGASE)<br>(ACYL- ACTIVATING<br>ENZYME) (ACETYL-COA<br>SYNTHASE) - BACILLUS<br>SUBTILIS, 572<br>aa. bcl3;SWISSPROT-ID:P39062<br>ACETYL-COENZYME A<br>SYNTHETASE (EC 6.2.1.1)<br>(ACETATE-COA LIGASE)<br>(ACYL- ACTIVATING<br>ENZYME) (ACETYL-COA<br>SYNTHASE) - BACILLUS<br>SUBTILIS, 572 aa. | 7.70E-79 | 16 |
| 455 | cg43124627 | 869  | TGGAAACAAGTGGAA<br>TATCCGAAAATGIA<br>/TCTGCACACACCCC<br>ACAGCAGTTGG | A<br>T           | Thr<br>(684)        | CONSER<br>VATIVE | synthase | Human Gene Similar to<br>SWISSNEW-ID:P39062<br>ACETYL-COENZYME A<br>SYNTHETASE (EC 6.2.1.1)<br>(ACETATE-COA LIGASE)<br>(ACYL- ACTIVATING<br>ENZYME) (ACETYL-COA<br>SYNTHASE) - BACILLUS<br>SUBTILIS, 572<br>aa. bcl3;SWISSPROT-ID:P39062<br>ACETYL-COENZYME A<br>SYNTHETASE (EC 6.2.1.1)<br>(ACETATE-COA LIGASE)<br>(ACYL- ACTIVATING<br>ENZYME) (ACETYL-COA<br>SYNTHASE) - BACILLUS<br>SUBTILIS, 572 aa. | 7.70E-79 | 16 |

|     |            |      |  |        |              |              |                  |              |  |           |
|-----|------------|------|--|--------|--------------|--------------|------------------|--------------|--|-----------|
| 456 | cg43064068 | 1464 | AGGAGAGGTGGTG<br>AAGGCCATTGTGIG<br>(ATCCCTGGCCTCGC<br>AGTTCCTGTCCCCA | G<br>A | Val<br>(685) | Ile<br>(685) | CONSER<br>VATIVE | synthase     | Human Gene Similar to<br>SWISSNEW-ID:P39062<br>ACETYL-COENZYME A<br>SYNTHETASE (EC 6.2.1.1)<br>(ACETATE-COA LIGASE)<br>(ACYL- ACTIVATING<br>ENZYME) (ACETYL-COA<br>SYNTHASE) - BACILLUS<br>SUBTILIS, 572<br>aa. b c;s:SWISSPROT-ID:P39062<br>ACETYL-COENZYME A<br>SYNTHETASE (EC 6.2.1.1)<br>(ACETATE-COA LIGASE)<br>(ACYL- ACTIVATING<br>ENZYME) (ACETYL-COA<br>SYNTHASE) - BACILLUS<br>SUBTILIS, 572 aa. | 7.40E-65  |
| 457 | cg2514276  | 1090 | GTGATGGACCCTC<br>TCATATATGCCTTA<br>TJCCGCAGCCAAGA<br>GATGCGGAAGACC   | A<br>T | Tyr<br>(686) | Phe<br>(686) | CONSER<br>VATIVE | tm7          | Human Gene SWISSPROT-<br>ID:P33032 MELANOCORTIN-5<br>RECEPTOR (MC5-R) (MC-2)-<br>HOMO SAPIENS (HUMAN), 325<br>aa.  | 7.00E-172 |
| 458 | cg22423505 | 964  | TTCATCTGAGGT<br>TATAAACACACG[A/<br>T]ATTCAAGCAAAG<br>TGGCCAGAATGGC   | A<br>T | Phe<br>(687) | Tyr<br>(687) | CONSER<br>VATIVE | tm7          | Human Gene Similar to<br>SPTREMBL-ID:Q8S609 G<br>PROTEIN-COUPLED<br>RECEPTOR - EQUINE<br>HERPESVIRUS TYPE 2 (EHV-2),<br>383 aa.  | 1.20E-55  |
| 459 | cg4335558  | 344  | CAAGACCTAGCTC<br>CCCAGCAGAGAGI<br>CTGGCCCCACAA<br>CAAAGAGGTCCA<br>GC | C<br>T | Ala<br>(688) | Val<br>(688) | CONSER<br>VATIVE | tnf receptor | Human Gene Similar to<br>TREMBL NEW-ID:Q2653845 TNF<br>RECEPTOR-RELATED<br>RECEPTOR FOR TRAIL - HOMO<br>SAPIENS (HUMAN), 386 aa.   | 5.50E-89  |

|     |            |      |   |   |   |              |              |                  |                  |   |           |          |
|-----|------------|------|---|---|---|--------------|--------------|------------------|------------------|---|-----------|----------|
| 460 | cg43998970 | 1347 | GACAGAGCTGTAC<br>CGTACATTTTC/C/<br>GJAGCACCTTCGGG<br>ATGAAATCAGGCAA | C | G | Gln<br>(689) | Glu<br>(689) | CONSER<br>VATIVE | transcriptfactor | Human Gene SPTREMBL-<br>ID:Q07279 TRANSCRIPTION<br>FACTOR NF-E2 - MUS<br>MUSCULUS (MOUSE), 373 aa.  | 1.70E-177 | 12       |
| 461 | cg2537639  | 800  | GAGGGCGATTCT<br>ACTACCTGGGG[G<br>[C]GTTCTCGGGGG<br>GTCGGGTGCAAGAG   | G | C | Gly<br>(690) | Ala<br>(690) | CONSER<br>VATIVE | transferase      | Human Gene SWISSPROT-<br>ID:PI6442<br>FUCOSYLYCOPROTEIN<br>ALPHA-N-<br>ACETYLGLACTOSAMINYLT<br>RANSFERASE (EC 2.4.1.40)<br>(HISTO-BLOOD GROUP A<br>TRANSFERASE) (A<br>TRANSFERASE) /<br>FUCOSYLYCOPROTEIN 3-<br>ALPHA-<br>GALACTOSYLTRANSFERASE<br>(EC 2.4.1.37) (HISTO-BLOOD<br>GROUP B TRANSFERASE) (B<br>TRANSFERASE) (NAGAT)-<br>HOMO SAPIENS (HUMAN), 354<br>aa. | 6.50E-192 | 9 (9q34) |

|     |            |      |  |   |   |              |              |                  |                  |   |   |               |
|-----|------------|------|--|---|---|--------------|--------------|------------------|------------------|---|---|---------------|
| 462 | cg43935995 | 1552 | AGTGTCCCCTCACC<br>ATGGTCACCCCTGIA<br>TGTCACCCCTGCCCT<br>TGCTTTCTCTCT | A | G | Ile          | Val<br>(691) | CONSER<br>VATIVE | transport        | Human Gene SWISSPROT-<br>ID:Q03518 ANTIGEN PEPTIDE<br>(PEPTIDE TRANSPORTER<br>TAP1) (PEPTIDE<br>TRANSPORTER PSF1)<br>(PEPTIDE SUPPLY FACTOR 1)<br>(PSF-1) (PEPTIDE<br>TRANSPORTER INVOLVED IN<br>ANTIGEN PROCESSING 1)-<br>HOMO SAPIENS (HUMAN), 748<br>aa. | 0 | 6             |
| 463 | cg43935986 | 1424 | CCTGGAACGGGCC<br>TTGTACCTGCTCTG/<br>ATAAGGAGGGTG<br>CTGCACTGGGG<br>T | G | A | Val<br>(692) | Ile<br>(692) | CONSER<br>VATIVE | transport        | Human Gene SPTREMBL-<br>ID:Q28437 ABC-TRANSPORTER<br>-GORILLA GORILLA GORILLA<br>(LOWLAND GORILLA), 703 aa.   | 0 | 6<br>(6p21.3) |
| 464 | cg43968274 | 730  | GAGCACGAGGAAG<br>CCATGAATGGGG[C<br>T]CTACTCAGGCTA<br>CGTCTACACGGCAC  | C | T | Ala          | Val<br>(693) | CONSER<br>VATIVE | UNCLASSIFI<br>ED | Human Gene SPTREMBL-<br>ACC:O14914 NEURONAL<br>MUNC18-1 BINDING PROTEIN-<br>HOMO SAPIENS (HUMAN), 837<br>aa.  | 0 | 9             |
| 465 | cg44018598 | 3568 | AGATACTTCTATA<br>AGCAGTTTTA[AG/C<br>TATTGTAGGAAGCA<br>GCTGAATTCAA    | G | C | Leu          | Val<br>(694) | CONSER<br>VATIVE | UNCLASSIFI<br>ED | Human Gene SWISSPROT-<br>ACC:P29374<br>RETINOBLASTOMA BINDING<br>PROTEIN 1 (RBPF-1) - Homo<br>sapiens (Human), 1257 aa.   | 0 | 14            |

|     |            |      |  |   |   |              |              |                  |                   |   |           |                     |
|-----|------------|------|--|---|---|--------------|--------------|------------------|-------------------|---|-----------|---------------------|
| 466 | cg44926796 | 1825 | ACACTGGAAAGCA<br>CAACAGTGGCAIC<br>(GTTCTGTCTAGAA<br>AATAATAATTGCA        | C | G | Thr<br>(695) | Ser<br>(695) | CONSER<br>VATIVE | UNCLASSIFIIE<br>D | Human Gene SWISSPROT-<br>ACC:Q15046 LYSYL-TRNA<br>SYNTHETASE (EC 6.1.1.6)<br>(LYSINE--TRNA LIGASE)<br>(LYSRS) (KIAA0070) - Homo<br>sapiens (Human), 597 aa. | 0         | 16                  |
| 467 | cg43055918 | 1622 | AACGCTGCCCTGA<br>CTGAGAAAGGCAI<br>CTTGATGGCTCGCTC<br>CACTGCTGGAAACC<br>G | C | T | Arg<br>(696) | His<br>(696) | CONSER<br>VATIVE | UNCLASSIFIIE<br>D | Human Gene SWISSPROT-<br>ACC:P42694 HYPOTHETICAL<br>PROTEIN KIAA0054 - Homo<br>sapiens (Human), 1942 aa.  | 0         | 17                  |
| 468 | cg43966985 | 1381 | CATCCAGGACAAC<br>TTCTCGGIGACTC/<br>GIAAGTGCCTTCA<br>CTGAGAGGCCCTG        | C | G | Gln<br>(697) | Glu<br>(697) | CONSER<br>VATIVE | UNCLASSIFIIE<br>D | Human Gene SWISSPROT-<br>ACC:P01019 ANGIOTENSINogen<br>PRECURSOR - Homo sapiens<br>(Human), 485 aa.   | 3.90E-257 | 1 (1q42)            |
| 469 | cg43918854 | 966  | CITCAACCCTGGIC<br>GGAGACACGGGA/<br>CTCACCATGGCCA<br>TCAGAACAGTGGCG       | A | C | Ile<br>(698) | Leu<br>(698) | CONSER<br>VATIVE | UNCLASSIFIIE<br>D | Human Gene SWISSPROT-<br>ACC:P20062 TRANSCOBALAMIN II<br>PRECURSOR - Homo sapiens<br>(Human), 427 aa.   | 3.30E-228 | 22<br>(22q11.2<br>) |
| 470 | cg43918484 | 1148 | CTGATTCTCCGTT<br>CTTCTTGACTTICG<br>TGCCACCTTGCCA<br>GCCAGCTGCTCG         | C | G | Glu<br>(699) | Gln<br>(699) | CONSER<br>VATIVE | UNCLASSIFIIE<br>D | Human Gene SWISSPROT-<br>ACC:P05089 ARGINASE 1 (EC<br>3.5.3.1) (LIVER-TYPE<br>ARGINASE) - Homo sapiens<br>(Human), 322 aa.                                  | 1.30E-171 | 6 (6q23)            |

|     |            |      |   |             |                            |                                     |                  |  |           |
|-----|------------|------|---|-------------|----------------------------|-------------------------------------|------------------|--|-----------|
| 471 | cg43942977 | 1009 | ACGGCCCTGGAGA<br>ACCAGAAAGAAGGI<br>C/TGAGGAAGAAGAG<br>AAAGTCTTGATTG<br>CC | C<br>T<br>T | Ala<br>Val<br>Asp<br>(700) | CONSER<br>VATIVE<br>VATIVE<br>(701) | UNCLASSIFI<br>ED | Human Gene Homologous to<br>SWISSNEW-ACC:Q12846<br>SYNTAXIN 4 - Homo sapiens<br>(Human), 297 aa.   | 9.60E-148 |
| 472 | cg43942977 | 725  | GGATGGTGTCTGA<br>TGAGGAGTTGGAG<br>TTCAGATGCTGGA<br>CAGTGGCAAAGC<br>G      | G<br>T<br>G | Glu<br>Asp<br>(701)        | CONSER<br>VATIVE<br>VATIVE<br>(702) | UNCLASSIFI<br>ED | Human Gene Homologous to<br>SWISSNEW-ACC:Q12846<br>SYNTAXIN 4 - Homo sapiens<br>(Human), 297 aa.   | 9.60E-148 |
| 473 | cg43943361 | 921  | TTGGGGTGGCTTG<br>GTTTCAATAAG[G/<br>C]AACGGGGACAT<br>TACAAATTGCTGC         | G<br>C      | Glu<br>Gln<br>(700)        | CONSER<br>VATIVE<br>VATIVE<br>(702) | UNCLASSIFI<br>ED | Human Gene Homologous to<br>SWISSNEW-ACC:P04179<br>SUPEROXIDE DISMUTASE<br>[Mn] PRECURSOR (EC 1.15.1.1)<br>- Homo sapiens (Human), 222 aa.   | 5.70E-124 |
| 474 | cg2236776  | 1094 | GTGACCGAGCCG<br>AGTGCCGCGAGG<br>G/TCTTTCACCGCC<br>GCGCCCCGCCAG<br>C       | G<br>T<br>T | Gly<br>Val<br>Gly<br>(703) | CONSER<br>VATIVE<br>VATIVE<br>(703) | UNCLASSIFI<br>ED | Human Gene Similar to<br>SWISSNEW-ACC:P01185<br>VASOPRESSIN-NEUROPHYSIN<br>2-COPEPTIN PRECURSOR<br>[CONTAINS: ARG-<br>VASOPRESSIN; NEUROPHYSIN<br>2 (NEUROPHYSIN-I);<br>COPEPTIN] - Homo sapiens<br>(Human), 164 aa. | 7.20E-91  |

|     |            |     |  |                               |                       |                 |   |          |
|-----|------------|-----|--|-------------------------------|-----------------------|-----------------|---|----------|
| 475 | cg25236776 | 881 | CAGTGCCTCCCTG<br>CGCCCCGGGG<br>TCAAAGCCGCTG<br>CITGGGCCAGC           | G<br>T<br>Gly<br>Val<br>(704) | CONSER<br>VATIVE<br>D | UNCLASSIFI<br>E | Human Gene Similar to<br>SWISSNEW-ACC:P01185<br>VASOPRESSIN-NEUROPHYSIN<br>2-COPEPTIN PRECURSOR<br>[CONTAINS: ARG-<br>VASOPRESSIN; NEUROPHYSIN<br>2 (NEUROPHYSIN-II);<br>COPEPTIN] - Homo sapiens<br>(Human), 164 aa. | 7.20E-91 |
| 476 | cg38899722 | 30  | GGCCAACCTCTGCT<br>ATGGACACCAAGA<br>G/C/TACTCTGCTGT<br>GGGGTCATCTGTCT | G<br>C<br>Val<br>Leu<br>(705) | CONSER<br>VATIVE<br>D | UNCLASSIFI<br>E | Human Gene Similar to<br>REMTREMBL-ACC:G292791 T-<br>CELL RECEPTOR BETA<br>PRECURSOR - HOMO SAPIENS<br>(HUMAN), 145 aa (fragment).  | 5.70E-75 |
| 477 | cg11753818 | 253 | GCCTGGAACACCCA<br>GGCTCCCTGCCIG/<br>ATGGTCATGCTTIG<br>TCTCCTGGGAGCA  | G<br>A<br>Arg<br>His<br>(706) | CONSER<br>VATIVE<br>D | UNCLASSIFI<br>E | Human Gene Similar to<br>REMTREMBL-ACC:G2104755 T<br>CELL RECEPTOR V-BETA 23 -<br>HOMO SAPIENS (HUMAN), 129<br>aa (fragment).   | 1.30E-66 |
| 478 | cg2526759  | 519 | AGCCACCCAGACC<br>GGAGACTCTGGCCIG<br>ATCTACCTCTGTG<br>CTGGGAGGCCAA    | G<br>A<br>Val<br>Ile<br>(707) | CONSER<br>VATIVE<br>D | UNCLASSIFI<br>E | Human Gene Similar to<br>REMTREMBL-ACC:G33509 T<br>CELL RECEPTOR - HOMO<br>SAPIENS (HUMAN), 118 aa<br>(fragment).   | 1.60E-54 |
| 479 | cg2526759  | 539 | CGCCCGTCTACCTC<br>TGTCCTGTGGAIG/<br>CJGCCTATCTAAC<br>GAATACAAGCTCA   | G<br>C<br>Glu<br>Asp<br>(708) | CONSER<br>VATIVE<br>D | UNCLASSIFI<br>E | Human Gene Similar to<br>REMTREMBL-ACC:G33509 T<br>CELL RECEPTOR - HOMO<br>SAPIENS (HUMAN), 118 aa<br>(fragment).   | 1.60E-54 |

|     |            |      |   |   |   |     |              |                          |                       |  |                    |
|-----|------------|------|---|---|---|-----|--------------|--------------------------|-----------------------|--|--------------------|
| 480 | cg1902363  | 368  | CAGAACAAAGCA<br>AATGGAATTGGAG<br>TJAGCATCCTGGTG<br>GCCCTGCTGCAGA        | G | T | Glu | Asp<br>(709) | CONSER<br>VATIVE         | UNCLASSIFI<br>ED      | Human Gene Similar to<br>SWISSPROT-ACC:P01286<br>SOMATOLIBERIN PRECURSOR<br>(GROWTH HORMONE-<br>RELEASING FACTOR) (GRF)<br>(GROWTH HORMONE-<br>RELEASING HORMONIF)<br>(GHRH) (SOMATOCRININ) -<br>Homo sapiens (Human), 108 aa. | 2.10E-52           |
| 481 | cg43277632 | 3110 | GAAACCCGGAAGC<br>ACTGTAATTGCG[A<br>/G]GGTCTATAAAT<br>GCACATGGCTCTG<br>T | A | G | Arg | Gly<br>(710) | NON-<br>CONSER<br>VATIVE | ATPase_associat<br>ed | Human Gene SWISSPROT-<br>ID:P35670 COPPER-<br>TRANSPORTING ATPASE 2 (EC<br>3.6.1.36) (COPPER PUMP 2)<br>(WILSON DISEASE-<br>ASSOCIATED PROTEIN) -<br>HOMO SAPIENS (HUMAN),<br>1465 aa.   | 0<br>(13q14.3<br>) |

|     |            |      |   |   |   |              |              |                           |                        |   |           |                      |
|-----|------------|------|---|---|---|--------------|--------------|---------------------------|------------------------|---|-----------|----------------------|
| 482 | cg43252813 | 2306 | TGTATTCCCTGTAAT<br>GGGGCTGATGA[C/<br>T]ATATATGATGGT<br>TATGGACCACAC | C | T | Thr<br>(711) | Ile<br>(711) | NON-<br>CONSER-<br>VATIVE | ATPase_associat-<br>ed | Human Gene SWISSNEW-<br>ID:Q04656 COPPER-<br>TRANSPORTING ATPASE 1 (EC<br>3.6.1.36) (COPPER PUMP 1)<br>(MENKES DISEASE-<br>ASSOCIATED PROTEIN)-<br>HOMO SAPIENS (HUMAN),<br>1500 aa, Ipcis:SWISSPROT-<br>ID:Q04656 COPPER-<br>TRANSPORTING ATPASE 1 (EC<br>3.6.1.36) (COPPER PUMP 1)<br>(MENKES DISEASE-<br>ASSOCIATED PROTEIN)-<br>HOMO SAPIENS (HUMAN),<br>1500 aa. | 0         | X (Xq12)             |
| 483 | cg43920913 | 929  | GCCCCTGAGGAGT<br>CAGGACCCGGCTC<br>/TICCGTCCGTGAGT<br>GCCACGATCCCAG  | C | T | Pro          | Ser<br>(712) | NON_<br>CONSER-<br>VATIVE | biotindep              | Human Gene SWISSPROT-<br>ID:P05166 PROPIONYL-COA<br>CARBOXYLASE BETA CHAIN<br>PRECURSOR (EC 6.4.1.3)<br>(PCCASE) (PROPANOYL-<br>COA:CARBON DIOXIDE<br>LIGASE) - HOMO SAPIENS<br>(HUMAN), 539 aa.  | 8.20E-288 | 3 (3q21)             |
| 484 | cg40310734 | 267  | GGAGTGGGTGCTG<br>CTGCTCTTGGGA[C/<br>G]CTTGTGCTGGCC<br>CTCCAGGCTGGGC | C | G | Pro          | Ala<br>(713) | NON-<br>CONSER-<br>VATIVE | cadherin               | Human Gene SWISSPROT-<br>ID:P08514 PLATELET<br>MEMBRANE GLYCOPROTEIN<br>IIb PRECURSOR (GP1IB)<br>(INTEGRIN ALPHA-IIb) (CD41)<br>- HOMO SAPIENS (HUMAN),<br>1039 aa.   | 0         | 17<br>(17q21.3<br>2) |

|     |            |      |  |   |   |              |              |                           |          |  |           |                      |
|-----|------------|------|--|---|---|--------------|--------------|---------------------------|----------|--|-----------|----------------------|
| 485 | cg40310734 | 3111 | CGTGTCCCTCCCTCC<br>CCTATGCCGTGIC/<br>GCCCCGGCTCAGCC<br>TGCCCCGAGGGGA | C | G | Pro<br>(714) | Ala<br>(714) | NON-<br>CONSER-<br>VATIVE | cadherin | Human Gene SWISSPROT-<br>ID:P08514 PLATELET<br>MEMBRANE GLYCOPROTEIN<br>IIb PRECURSOR (GP1b)<br>(INTEGRIN ALPHA-IIb) (CD41)<br>-HOMO SAPIENS (HUMAN),<br>1039 aa.  | 0         | 17<br>(17q21.3<br>2) |
| 486 | cg43956560 | 777  | GGGGTACTATGGG<br>CCCCAGTGTCA GT/<br>CJTGTGATTCA GT/<br>GTGAGCCCTTGGA | T | C | Phe<br>(715) | Leu<br>(715) | NON-<br>CONSER-<br>VATIVE | cadherin | Human Gene SWISSPROT-<br>ID:P14151 L-SELECTIN<br>PRECURSOR (LYMPH NODE<br>HOMING RECEPTOR)<br>(LEUKOCYTE ADHESION<br>MOLECULE-1) (LAM-1)<br>(LEUKOCYTE SURFACE<br>ANTIGEN LEU-8) (TQ1) (GP90-<br>MEL) (LEUKOCYTE-<br>ENDOTHELIAL CELL<br>ADHESION MOLECULE 1)<br>(LECAM1) (CD62L) - HOMO<br>SAPIENS (HUMAN), 372 aa. | 1.00E-218 | 1 (1q23)             |

|     |            |      |  |   |   |              |              |                           |                 |  |           |          |
|-----|------------|------|--|---|---|--------------|--------------|---------------------------|-----------------|--|-----------|----------|
| 487 | cg4395660  | 837  | GCTGGGTACCATG<br>GAATGTACTCAC/C/<br>TCTTGGAAACT<br>TCAGCTTAGCTC          | C | T | Pro<br>(716) | Ser<br>(716) | NON-<br>CONSER-<br>VATIVE | cadherin        | Human Gene SWISSPROT-<br>ID:P14151L-SELECTIN<br>PRECURSOR (LYMPH NODE<br>HOMING RECEPTOR)<br>(LEUKOCYTE ADHESION<br>MOLECULE-1) (LAM-1)<br>(LEUKOCYTE SURFACE<br>ANTIGEN LEU-8) (CTQ1) (GP90-<br>MEL) (LEUKOCYTE-<br>ENDOTHELIAL CELL<br>ADHESION MOLECULE 1)<br>(LECAM1) (CD62L) - HOMO<br>SAPIENS (HUMAN), 372 aa. | 1.00E-218 | 1 (1q23) |
| 488 | cg4238809  | 753  | TGCAGAAGGCCACC<br>ACAGAGACCCGGAA<br>A/GGGCAGGGCAA<br>GGGCACCTCGAAC<br>AC | A | G | Arg<br>(717) | Gly<br>(717) | NON-<br>CONSER-<br>VATIVE | cadherin        | Human Gene SWISSPROT-<br>ID:P21815 BONE<br>SIALOPROTEIN II PRECURSOR<br>(BSP II) (CELL-BINDING<br>SIALOPROTEIN) (INTEGRIN-<br>BINDING SIALOPROTEIN)-<br>HOMO SAPIENS (HUMAN), 317<br>aa.   | 7.00E-172 | 4        |
| 489 | cg43977436 | 1945 | GTTGTGTGTAAT<br>GGTGTGGCTGTAIC<br>/TGTGCTCCAACCAA<br>GATCTTATTACTGA      | C | T | Arg<br>(718) | Cys<br>(718) | NON-<br>CONSER-<br>VATIVE | calcium_channel | Human Gene SWISSPROT-<br>ID:P21817 RYANODINE<br>RECEPTOR, SKELETAL<br>MUSCLE (SKELETAL MUSCLE<br>CALCIUM RELEASE<br>CHANNEL) - HOMO SAPIENS<br>(HUMAN), 5032 aa.   | 0         |          |

|     |             |      |  |   |   |     |              |                           |             |   |   |         |
|-----|-------------|------|--|---|---|-----|--------------|---------------------------|-------------|---|---|---------|
| 490 | cgt3280376  | 1130 | CGGAAGCTGGTGT<br>CCTACTGCCCA/<br>GAAAGGTGCAACA<br>ACTGTTGCCCTC       | A | G | Gln | Arg<br>(719) | NON-<br>CONSER-<br>VATIVE | carboxylase | Human Gene SWISSPROT-<br>ID:P38435 VITAMIN K-<br>CARBOXYLASE (EC 6.4.-.)<br>(GAMMA-GLUTAMYL<br>CARBOXYLASE) - HOMO<br>SAPIENS (HUMAN), 758 aa | 0 | 2       |
| 491 | cgt42201364 | 1595 | CCAGGGCCTCCAG<br>GTCCAAGAGGCCA/<br>C/CCTGGAGAGC<br>CTGGTCTTCAGG<br>G | A | C | Tyr | Gly<br>(720) | NON-<br>CONSER-<br>VATIVE | collagen    | Human Gene SWISSPROT-<br>ID:Q03692 COLLAGEN ALPHA<br>I(X) CHAIN PRECURSOR -<br>HOMO SAPIENS (HUMAN), 680<br>aa.                               | 0 | 6       |
| 492 | cgt42201364 | 176  | GTTTTACGCTGA<br>ACGATACAAAC/<br>TJGCCAACAGGCAT<br>AAAAGGCCACTA       | C | T | Thr | Met<br>(721) | NON-<br>CONSER-<br>VATIVE | collagen    | Human Gene SWISSPROT-<br>ID:Q03692 COLLAGEN ALPHA<br>I(X) CHAIN PRECURSOR -<br>HOMO SAPIENS (HUMAN), 680<br>aa.                               | 0 | 6       |
| 493 | cgt40339378 | 2855 | TCCAGGAATACCA<br>GGTCTGGCTGGT/A/<br>GTTCCCTGGAACAA<br>GAGGATTAAAGG   | A | G | Ile | Thr<br>(722) | NON-<br>CONSER-<br>VATIVE | collagen    | Human Gene SPTREMBL-<br>ID:Q12823 A TYPE IV<br>COLLAGEN - HOMO SAPIENS<br>(HUMAN), 1690 aa (fragment).  | 0 | X(Xq22) |

|     |            |      |   |   |   |              |              |                           |            |   |           |          |
|-----|------------|------|---|---|---|--------------|--------------|---------------------------|------------|---|-----------|----------|
| 494 | cg43063256 | 606  | AAGACTGTGTTACC<br>AACAGACCATGCGA<br>/G]GAAGTCAAAGTG<br>CGATGTGAAGGCT<br>T | A | G | Arg<br>(723) | Gly<br>(723) | NON-<br>CONSER-<br>VATIVE | complement | Human Gene SWISSPROT-<br>ID:P07338 COMPLEMENT<br>COMPONENT C8 BETA CHAIN<br>PRECURSOR - HOMO SAPIENS<br>(HUMAN), 591<br>aa.[pels;SWISSPROT-ID:P07338<br>COMPLEMENT COMPONENT<br>C8 BETA CHAIN PRECURSOR -<br>HOMO SAPIENS (HUMAN), 591<br>aa. | 0         | 1 (1p32) |
| 495 | cg44032748 | 414  | CTCCAGTTCTACAA<br>CTTGTGTAAGGIA/<br>CJAAGGCACAGTGTG<br>GACAGGGATTCCA      | A | C | Lys<br>(724) | Gln<br>(724) | NON-<br>CONSER-<br>VATIVE | complement | Human Gene SWISSPROT-<br>ID:P07357 COMPLEMENT<br>COMPONENT C8 ALPHA<br>CHAIN PRECURSOR - HOMO<br>SAPIENS (HUMAN), 584 aa.   | 0         | 1 (1p32) |
| 496 | cg43049885 | 533  | CAGTTGGGGAC<br>AGCCATGCACTGIA<br>/C]GCCCTCTGGTAGC<br>CTTCAACCATGC         | A | C | Glu<br>(725) | Ala<br>(725) | NON-<br>CONSER-<br>VATIVE | complement | Human Gene TREMBLNEW-<br>ID:G386348 COMPLEMENT C6 -<br>HOMO SAPIENS, 941 aa.  | 0         | 5 (5p13) |
| 497 | cg2164442  | 1347 | CCAGGGCTCTCCCCA<br>GGATCTCATCAC[T/<br>C]GGGGCCCCAGGG<br>CCTCAGCAACCCC     | T | C | Leu<br>(726) | Pro<br>(726) | NON-<br>CONSER-<br>VATIVE | csf        | Human Gene SWISSPROT-<br>ID:P09663 MACROPHAGE<br>COLONY STIMULATING<br>FACTOR-1 PRECURSOR (CSF-1)<br>(M-CSF) - HOMO SAPIENS<br>(HUMAN), 554 aa.   | 5.00E-304 | 1 (1p21) |

|     |            |      |   |   |   |     |              |                           |            |   |          |    |
|-----|------------|------|---|---|---|-----|--------------|---------------------------|------------|---|----------|----|
| 498 | cg7753430  | 279  | CCAAGCTCCCCATG<br>ACCCAGACAACGIC<br>TTCCTGAAGACA<br>AGCTGGTTAACT<br>G | C | T | Pro | Ser<br>(727) | NON-<br>CONSER-<br>VATIVE | csf        | Human Gene Similar to<br>SWISSNEW-ID:P08700<br>INTERLEUKIN-3 PRECURSOR<br>(IL-3) (MULTIPOTENTIAL<br>COLONY-STIMULATING<br>FACTOR) (HEMATOPOIETIC<br>GROWTH FACTOR) (P-CELL<br>STIMULATING FACTOR)<br>(MAST-CELL GROWTH<br>FACTOR) (MCGF) - HOMO<br>SAPIENS (HUMAN), 152<br>aa, Ipcds, SWISSPROT-ID:P08700<br>INTERLEUKIN-3 PRECURSOR<br>(IL-3) (MULTIPOTENTIAL<br>COLONY-STIMULATING<br>FACTOR) (HEMATOPOIETIC<br>GROWTH FACTOR) (P-CELL<br>STIMULATING FACTOR)<br>(MAST-CELL GROWTH<br>FACTOR) (MCGF) - HOMO<br>SAPIENS (HUMAN), 152 aa. | 1.10E-77 | 5  |
| 499 | cg73923204 | 1651 |   | A | G | Tyr | His<br>(728) | NON-<br>CONSER-<br>VATIVE | cytochrome | Human Gene Similar to<br>SWISSPROT-ID:P21592<br>CYTOCHROME C OXIDASE<br>ASSEMBLY PROTEIN COX10<br>PRECURSOR -<br>SACCHAROMYCES<br>CEREVISIAE (BAKER'S<br>YEAST), 462 aa.  | 1.70E-52 | 17 |

|     |            |      |  |   |   |              |              |                           |               |  |           |               |
|-----|------------|------|--|---|---|--------------|--------------|---------------------------|---------------|--|-----------|---------------|
| 500 | cg44017721 | 174  | TGGTAGGGACGG<br>AACTCGGGCGC[G<br>T]GGCGGTGGCC<br>GAGTGGAGATAGG<br>A      | G | T | Pro<br>(729) | Gln<br>(729) | NON-<br>CONSER-<br>VATIVE | cytochrome    | Human Gene Similar to<br>SPTREMBL-ID:000761<br>CYTOCHROME OXIDASE<br>SUBUNIT VIA HEART<br>ISOFORM PRECURSOR (EC<br>1.9.3.1) (CYTOCHROME-C<br>OXIDASE) (CYTOCHROME<br>A(3)) (CYTOCHROME AA(3))<br>-<br>HOMO SAPIENS (HUMAN), 97<br>aa.                            | 2.40E-52  | 22            |
| 501 | cg11626024 | 279  | GCTGGTTTGCTCCC<br>AGGAGGCCAAG/A/<br>CAGTCAGCCTACT<br>GCCCTAACAGTCA       | A | C | Lys<br>(730) | Gln<br>(730) | NON-<br>CONSER-<br>VATIVE | deaminase     | Human Gene Similar to<br>SWISSPROT-ID:P32320<br>CYTIDINE DEAMINASE (EC<br>3.5.4.5) (CYTIDINE<br>AMINOHYDROLASE) - HOMO<br>SAPIENS (HUMAN), 146<br>aa.[pcis:TREMBLNEW-<br>ID:E1228801 CYTIDINE<br>DEAMINASE (EC 3.5.4.5)<br>-<br>HOMO SAPIENS (HUMAN), 146<br>aa. | 8.80E-73  | 1<br>(Ip36.2) |
| 502 | cg43057018 | 1618 | AAGGCATCCGAACA<br>ATCCCTCATCTTTT/<br>G[GAAAGATGCCAG<br>GAGCAATTGGAA<br>T | T | G | End          | Gly<br>(731) | NON-<br>CONSER-<br>VATIVE | dehydrogenase | Human Gene SWISSNEW-<br>ID:P08319 ALCOHOL<br>DEHYDROGENASE CLASS II PI<br>CHAIN (EC 1.1.1.1) - HOMO<br>SAPIENS (HUMAN), 391<br>aa.[pcis:SWISSPROT-ID:P08319<br>ALCOHOL DEHYDROGENASE<br>CLASS II PI CHAIN (EC 1.1.1.1)<br>-<br>HOMO SAPIENS (HUMAN), 391<br>aa.  | 1.30E-209 | 4<br>(4q22)   |

|     |            |      |   |   |   |              |              |                           |             |  |          |                    |
|-----|------------|------|---|---|---|--------------|--------------|---------------------------|-------------|--|----------|--------------------|
| 503 | cgt2837709 | 464  | CGCACCAACGCC<br>GACATCATCGAGIA<br>G CCCTGAGGAAG<br>AAGGGCTCAAGG<br>G  | A | G | Thr<br>(732) | Ala<br>(732) | NON-<br>CONSER-<br>VATIVE | dna_ma_bind | Human Gene Similar to<br>TREMBLNEW-ID:G913312 DNA-<br>BINDING PROTEIN MEF2<br>{CLONE XMEF2A.1} -<br>XENOPUS LAEVIS; 516 aa.  | 3.90E-86 | 1                  |
| 504 | cgt3327954 | 2205 | TCCACGACGGGT<br>AGAGAACTACAA<br>C A]CCGGCGCAGC<br>GCAAGCTCCGCAA<br>CC | C | A | Asn<br>(733) | Lys<br>(733) | NON-<br>CONSER-<br>VATIVE | dna_ma_bind | Human Gene Similar to<br>SPTREMBL-ID:Q61491 DNA-<br>BINDING PROTEIN - MUS<br>MUSCULUS (MOUSE); 546 aa.   | 5.50E-57 | 1                  |
| 505 | cgt3971258 | 707  | TCGTTGGAGATGA<br>CAAGTCCGGAGIC<br>/T]GAGCTCGGCTGT<br>CTGGATGGAAAGG    | C | T | Ala<br>(734) | Thr<br>(734) | NON-<br>CONSER-<br>VATIVE | dna_ma_bind | Human Gene Similar to<br>SWISSNEW-ID:Q02535 DNA-<br>BINDING PROTEIN INHIBITOR<br>ID-3 (ID-LIKE PROTEIN<br>INHIBITOR HLH1R21) (HELIX-<br>LOOP-HELIX PROTEIN HEIR-1)<br>-HOMO SAPIENS (HUMAN),<br>119 aa.[pcl:SWISSPROT-<br>ID:Q02535 DNA-BINDING<br>PROTEIN INHIBITOR ID-3 (ID-<br>LIKE PROTEIN INHIBITOR<br>HLH IR21) (HELIX-LOOP-<br>HELIX PROTEIN HEIR-1)-<br>HOMO SAPIENS (HUMAN), 119<br>aa. | 1.30E-60 | 1<br>(1p36.13<br>) |

|     |            |      |   |                        |              |                          |          |  |                            |
|-----|------------|------|---|------------------------|--------------|--------------------------|----------|--|----------------------------|
| 506 | cg41554010 | 1253 | AGCTGGAGCAACA<br>GCAGGAAACAGCA<br>GT[CA]GGAGCAGC<br>AGCAGGAGCAGGT<br>GC | G<br>T<br>Gln<br>(735) | Hs<br>(735)  | NON-<br>CONSER-<br>ATIVE | eph      | Human Gene SWISSNEW-<br>ID:P06727 APOLIPOPROTEIN A-<br>IV PRECURSOR (APO-AIV) -<br>HOMO SAPIENS (HUMAN), 396<br>aa.[pcls:SWISSPROT-ID:P06727<br>APOLIPOPROTEIN A-IV<br>PRECURSOR (APO-AIV) -<br>HOMO SAPIENS (HUMAN), 396<br>aa.   | 1.80E-203<br>11<br>(11q23) |
| 507 | cg43957743 | 1063 | GTTGGCATACCTG<br>GATATTAA[TCT<br>TCAGTGAGATAA<br>AAGACAGCCACT           | C<br>T<br>Gly<br>(736) | Glu<br>(736) | NON-<br>CONSER-<br>ATIVE | esterase | Human Gene SWISSNEW-<br>ID:Q15166 SERUM<br>PARAOXONASE/ARYLESTERA-<br>SE 3 (EC 3.1.1.2) (EC 3.1.8.1)<br>(PON 3) (SERUM<br>ARYLDIAKYLPHOSPHATASE<br>3) (A-ESTERASE 3)<br>(AROMATIC ESTERASE 3)<br>HOMO SAPIENS (HUMAN), 341<br>aa (fragment).[pcls:SWISSPROT-<br>ID:Q15166 SERUM<br>PARAOXONASE/ARYLESTERA-<br>SE 3 (EC 3.1.1.2) (EC 3.1.8.1)<br>(PON 3) (SERUM<br>ARYLDIAKYLPHOSPHATASE<br>3) (A-ESTERASE 3)<br>(AROMATIC ESTERASE 3)<br>HOMO SAPIENS (HUMAN), 341<br>aa (fragment). | 1.90E-178                  |

|     |            |      |  |   |   |     |              |                           |          |  |           |               |
|-----|------------|------|--|---|---|-----|--------------|---------------------------|----------|--|-----------|---------------|
| 508 | cg43957743 | 1079 | TATTTAATCCAGT<br>GGAGATAAAAGIA/<br>CICAGCCCCACTAGG<br>AAGTATATCAATA    | A | C | Ser | Ala<br>(737) | NON-<br>CONSER-<br>VATIVE | esterase | Human Gene SWISSPROT-<br>ID:Q15166 SERUM<br>PARAOXONASE/ARYLESTERA-<br>SE 3 (EC 3.1.1.2) (EC 3.1.8.1)<br>(PON 3) (SERUM<br>ARYLDIACYLPHOSPHATASE<br>3) (A-ESTERASE 3)<br>(AROMATIC ESTERASE 3)-<br>HOMO SAPIENS (HUMAN), 341<br>aa (fragment). [pcis:SWISSPROT-<br>ID:Q15166 SERUM<br>PARAOXONASE/ARYLESTERA-<br>SE 3 (EC 3.1.1.2) (EC 3.1.8.1)<br>(PON 3) (SERUM<br>ARYLDIACYLPHOSPHATASE<br>3) (A-ESTERASE 3)<br>(AROMATIC ESTERASE 3)-<br>HOMO SAPIENS (HUMAN), 341<br>aa (fragment). | 1.90E-178 |               |
| 509 | cg43248101 | 812  | AAGTGAATCTAT<br>CTTGCAATGAACIA<br>/G]AGGAAGGAAAA<br>CTCTATGCAAAGA<br>A | A | G | Lys | Glu<br>(738) | NON-<br>CONSER-<br>VATIVE | fgf      | Human Gene Homologous to<br>SWISSPROT-ID:P21781<br>KERATINOCYTE GROWTH<br>FACTOR PRECURSOR (KGF)<br>(FIBROBLAST GROWTH<br>FACTOR-7) (FGF-7) (HBGF-7)-<br>HOMO SAPIENS (HUMAN), 194<br>aa.  | 9.30E-106 | 15<br>(15q15) |

|     |            |     |   |   |   |              |              |                           |               |   |          |    |
|-----|------------|-----|---|---|---|--------------|--------------|---------------------------|---------------|---|----------|----|
| 510 | cg43969014 | 332 | GATGAGCTCTCCA<br>ACACCGTAAATTTC<br>ATTCGTTTTGAT<br>CCAGACCCAGATG    | C | A | Arg<br>(739) | Ile<br>(739) | NON-<br>CONSER-<br>VATIVE | glucuronidase | Human Gene Similar to<br>SWISSPROT-ID:P08236 BETA-<br>GLUCURONIDASE<br>PRECURSOR (EC 3.2.1.31)<br>(BETA-G1) - HOMO SAPIENS<br>(HUMAN), 651 aa.  | 7.40E-80 | 5  |
| 511 | cg43286488 | 387 | CACCAAGCAAGATG<br>CCCACGATCAGCG<br>/CIGAACCTGCCCA<br>AGGCCTGCTTCTTG | G | C | Pro<br>(740) | Arg<br>(740) | NON-<br>CONSER-<br>VATIVE | glycoprotein  | Human Gene SWISSNEW-<br>ID:P40967 MELANOCYTE<br>PROTEIN PMEL 17<br>PRECURSOR (MELANOCYTE<br>LINEAGE-SPECIFIC ANTIGEN<br>GP100) (MELANOMA,<br>ASSOCIATED ME20 ANTIGEN)<br>(ME20M/ME20S) (ME20-<br>M/ME20-S) (95 KD<br>MELANOCYTE-SPECIFIC<br>SECRETED GLYCOPROTEIN)-<br>HOMO SAPIENS (HUMAN), 661<br>aa.;pcr;SWISSPROT-ID:P40967<br>MELANOCYTE PROTEIN PMEL<br>17 PRECURSOR<br>(MELANOCYTE LINEAGE-<br>SPECIFIC ANTIGEN GP100)<br>(MELANOMA ASSOCIATED<br>ME20 ANTIGEN) (ME20M)<br>(ME20-M/ ME20-S) (95 KD<br>MELANOCYTE-SPECIFIC<br>SECRETED GLYCOPROTEIN)-<br>HOMO SAPIENS (HUMAN), 661<br>aa. | 0        | 12 |

|     |            |     |  |   |   |              |              |                           |              |  |   |
|-----|------------|-----|--|---|---|--------------|--------------|---------------------------|--------------|--|---|
| 512 | cg44004239 | 663 | TITTCCCAGGGGT<br>CACAGACTGATA/<br>G]ACCCACAGGGT<br>CAGGGTCTCTGT      | A | G | Tyr<br>(741) | His<br>(741) | NON-<br>CONSER-<br>VATIVE | glycoprotein | Human Gene SWISSPROT-<br>ID:Q12889 OVIDUCT-SPECIFIC<br>GLYCOPROTEIN PRECURSOR<br>(OVIDUCTAL<br>GLYCOPROTEIN)<br>(OVIDUCTIN) (ESTROGEN-<br>DEPENDENT OVIDUCT<br>PROTEIN) - HOMO SAPIENS<br>(HUMAN), 678 aa. | 0 |
| 513 | cg44004239 | 672 | GGGGTCAAGACT<br>GATAACCACAGIA<br>(G)GGTCAGGGTCT<br>TCTGTCCAGGGTC     | A | G | Ser<br>(742) | Pro<br>(742) | NON-<br>CONSER-<br>VATIVE | glycoprotein | Human Gene SWISSPROT-<br>ID:Q12889 OVIDUCT-SPECIFIC<br>GLYCOPROTEIN PRECURSOR<br>(OVIDUCTAL<br>GLYCOPROTEIN)<br>(OVIDUCTIN) (ESTROGEN-<br>DEPENDENT OVIDUCT<br>PROTEIN) - HOMO SAPIENS<br>(HUMAN), 678 aa. | 0 |
| 514 | cg44004239 | 773 | CTGATGACCCACA<br>GAAGTCAATGGTCIA<br>(G)TTGCCCAAGTG<br>ATCTCAGTCCTCTC | A | G | Met          | Thr<br>(743) | NON-<br>CONSER-<br>VATIVE | glycoprotein | Human Gene SWISSPROT-<br>ID:Q12889 OVIDUCT-SPECIFIC<br>GLYCOPROTEIN PRECURSOR<br>(OVIDUCTAL<br>GLYCOPROTEIN)<br>(OVIDUCTIN) (ESTROGEN-<br>DEPENDENT OVIDUCT<br>PROTEIN) - HOMO SAPIENS<br>(HUMAN), 678 aa. | 0 |

|     |            |      |   |   |   |           |           |                  |              |  |           |             |
|-----|------------|------|---|---|---|-----------|-----------|------------------|--------------|--|-----------|-------------|
| 515 | cg43932434 | 1504 | ATATGIGTCACTACTGGAGGTGTTGIG/TATGTGACGAAIGTACACCCCTGTGTT | G | T | Ser (744) | Tyr (744) | NON-CONSERVATIVE | glycoprotein | Human Gene SWISSPROT-ID:P16070 CD44 ANTIGEN PRECURSOR (PHAGOCYTIC GLYCOPROTEIN I) (PGP-1) (HUTCH-I) (EXTRACELLULAR MATRIX RECEPTOR-III) (ECM-R-III) (GP90 LYMPHOCTYE HOMING/ADHESION RECEPTOR) (HERMES ANTIGEN) (HYALURONATE RECEPTOR) (HEPARAN SULFATE PROTEOGLYCAN) (EPICAN) (CDW44) - HOMO SAPIENS (HUMAN), 742 aa.   | 1.80E-195 | 11 (11pter) |
| 516 | cg40915005 | 622  | AAGGAGCCCTCTCTC/CCTTCCATGTCATC/TCTGGATGGCATCTTTACAACCAT | C | T | Thr (745) | Ile (745) | NON-CONSERVATIVE | glycoprotein | Human Gene SWISSNEW-ID:P06126 T-CELL SURFACE GLYCOPROTEIN CD1A PRECURSOR (CD1A ANTIGEN) (T-CELL SURFACE ANTIGEN T6/LEU-6) (HTA1 THYMOCYTE ANTIGEN) - HOMO SAPIENS (HUMAN), 327 aa. pcbs:SWISSPROT-ID:P06126 T-CELL SURFACE GLYCOPROTEIN CD1A PRECURSOR (CD1A ANTIGEN) (T-CELL SURFACE ANTIGEN T6/LEU-6) (HTA1 THYMOCYTE ANTIGEN) - HOMO SAPIENS (HUMAN), 327 aa. | 2.00E-183 | 1 (1q21)    |

|     |            |      |  |   |   |     |              |                  |              |  |           |          |
|-----|------------|------|--|---|---|-----|--------------|------------------|--------------|--|-----------|----------|
| 517 | cg40915005 | 737  | ATTCAGCACCATTGTTTCCCTGTGG/CJCCCTGGCCAGGGAAACTTCAGCA      | G | C | Trp | Cys<br>(746) | NON-CONSERVATIVE | glycoprotein | Human Gene SWISSNEW-ID:P06126 T-CELL SURFACE GLYCOPROTEIN CD1A PRECURSOR (CD1A ANTIGEN) (T-CELL SURFACE ANTIGEN T6/LEU-6) (HTA1 THYMOCYTE ANTIGEN) - HOMO SAPIENS (HUMAN), 327 aa.[pcds:SWISSPROT-ID:P06126 T-CELL SURFACE GLYCOPROTEIN CD1A PRECURSOR (CD1A ANTIGEN) (T-CELL SURFACE ANTIGEN T6/LEU-6) (HTA1 THYMOCYTE ANTIGEN) - HOMO SAPIENS (HUMAN), 327 aa. | 2.00E-183 | 1 (1q21) |
| 518 | cg36834323 | 1529 | GTGCTCCCTGATCCA/GTGTGAAGCAT[A/GTGTTAGCTCAAGTTATGTGGCATCT | A | G | Tyr | Cys<br>(747) | NON-CONSERVATIVE | glycoprotein | Human Gene Similar to SWISSPROT-ID:P38159 HETEROGENEOUS NUCLEAR RIBONUCLEOPROTEIN G (HNRNP G) (GLYCOPROTEIN P43) - HOMO SAPIENS (HUMAN), 437 aa.   | 6.40E-91  |          |
| 519 | cg36834323 | 329  | AATGCTGCCAAAGATATGAATGGAAACGTCCTTGATGGAAAGCAATAAAA       | A | C | Lys | Thr<br>(748) | NON-CONSERVATIVE | glycoprotein | Human Gene Similar to SWISSPROT-ID:P38159 HETEROGENEOUS NUCLEAR RIBONUCLEOPROTEIN G (HNRNP G) (GLYCOPROTEIN P43) - HOMO SAPIENS (HUMAN), 437 aa.   | 6.40E-91  |          |

|     |            |      |   |   |   |              |              |                          |              |  |           |    |
|-----|------------|------|---|---|---|--------------|--------------|--------------------------|--------------|--|-----------|----|
| 520 | cg36834323 | 463  | AAGTCTGAGATCT<br>GCAAGAGGAAGC<br>A/C/GTGGAGGAAC<br>AAGGGGGCCT<br>CC | A | C | Ser<br>(749) | Arg<br>(749) | NON-<br>CONSER<br>VATIVE | glycoprotein | Human Gene Similar to<br>SWISSPROT-ID:P38159<br>HETEROGENEOUS NUCLEAR<br>RIBONUCLEOPROTEIN G<br>(HNRNP G) (GLYCOPROTEIN<br>P43) - HOMO SAPIENS<br>(HUMAN), 437 aa.                                       | 6.40E-91  |    |
| 521 | cg44019290 | 1697 | GCGGATAAGTAGA<br>GGACCTTCATGTT<br>GIGTATTGCTGGT<br>GAAGGTGGTCGG     | T | G | Asn<br>(750) | His<br>(750) | NON-<br>CONSER<br>VATIVE | glycoprotein | Human Gene Similar to<br>SWISSPROT-ID:P04216 THY-1<br>MEMBRANE GLYCOPROTEIN<br>PRECURSOR (THY-1 ANTIGEN)<br>(CDW90) (CD90 ANTIGEN) -<br>HOMO SAPIENS (HUMAN), 161<br>aa.                                 | 2.50E-80  | 11 |
| 522 | cg42336656 | 1665 | CTTAGACATACAA<br>TATACTTACCTTA/<br>GJGAGGTCACTAT<br>GTTTGTCCGCACA   | A | G | Arg<br>(751) | Gly<br>(751) | NON-<br>CONSER<br>VATIVE | glycoprotein | Human Gene Similar to<br>SWISSPROT-ID:Q05910 CELL<br>SURFACE ANTIGEN MS2<br>PRECURSOR (EC 3.4.24.-)<br>(MACROPHAGE CYSTEINE-<br>RICH GLYCOPROTEIN) (CD156<br>ANTIGEN) - MUS MUSCULUS<br>(MOUSE), 826 aa. | 9.40E-58  |    |
| 523 | cg42730678 | 980  | GGAGCGAGCGTGG<br>ATCCAGTTGCGCG<br>TTCGGGGTTGTTTG<br>GGTCAAGTGCTG    | G | T | Ala<br>(752) | Asp<br>(752) | NON-<br>CONSER<br>VATIVE | homeobox     | Human Gene SWISSPROT-<br>ID:P28356 HOMEobox<br>PROTEIN HOX-D9 (HOX-4C)<br>(HOX-5.2) - HOMO SAPIENS<br>(HUMAN), 342 aa.   | 2.60E-188 | 2  |

|     |            |      |   |   |   |              |              |                           |            |   |           |          |
|-----|------------|------|---|---|---|--------------|--------------|---------------------------|------------|---|-----------|----------|
| 524 | cg42714160 | 769  | GCCCTGTGCCCTGA<br>CGGAGAGGCAGA <br>T/GICAAGATAATGG<br>TICCAGAACCGAC<br>GC | T | G | Ile<br>(753) | Ser<br>(753) | NON-<br>CONSER-<br>VATIVE | homeobox   | Human Gene Homologous to<br>SWISSPROT-ID:P17509<br>HOMEBOX PROTEIN HOX-B6<br>(HOX-2B) (HOX-2.2) (HUMAN)-<br>HOMO SAPIENS (HUMAN), 224<br>aa.  | 1.10E-123 |          |
| 525 | cg42359655 | 3297 | CTGGGCACCATAT<br>AGGATAGGCCACIA<br>/G CCGTCAATCAA<br>GCCCATGCCAGAG<br>T   | A | G | Thr<br>(754) | Ala<br>(754) | NON-<br>CONSER-<br>VATIVE | hydrolase  | Human Gene SWISSPROT-<br>ID:P09848 LACTASE-<br>PHORVIN HYDROLASE<br>PRECURSOR (EC 3.2.1.108) (EC<br>3.2.1.62) (LACTASE-<br>GLYCOSYLCERAMIDASE)-<br>HOMO SAPIENS (HUMAN),<br>1927 aa.  | 0         | 2 (2q11) |
| 526 | cg43925670 | 2172 | GTGGAGGGTGAG<br>GTGAAGTAGCATIC<br>/GICACITTCITCTT<br>CCTCTTCTTGAT         | C | G | Asp<br>(755) | His<br>(755) | NON-<br>CONSER-<br>VATIVE | interferon | Human Gene SWISSPROT-<br>ID:Q16666 GAMMA-<br>INTERFERON-INDUCIBLE<br>PROTEIN IFI-16 (INTERFERON-<br>INDUCIBLE MYELOID<br>DIFFERENTIATION<br>TRANSCRIPTIONAL,<br>ACTIVATOR) - HOMO SAPIENS<br>(HUMAN), 729<br>aa. pcds:SPTRREMBL-ID:Q16666<br>IFI16=INTERFERON-<br>INDUCIBLE MYELOID<br>DIFFERENTIATION<br>TRANSCRIPTIONAL,<br>ACTIVATOR - HOMO SAPIENS<br>(HUMAN), 729 aa (fragment). | 0         | 1        |

| 527 | cg43090990 | 1083 | TGCTCCATCAAAA<br>ATGAAGCAAGGC<br>C/TGCCATGTTAC<br>CGACACCGGGAAA<br>A   | C | T | Pro<br>(756) | Leu<br>(756) | NON-<br>CONSER-<br>VATIVE | kinase | Human Gene SWISSPROT-<br>ID:Q04759 PROTEIN KINASE C,<br>THETA TYPE (EC 2.7.1.-)<br>(NPKC-THETA) - HOMO<br>SAPIENS (HUMAN), 706 aa.  | 0<br>10                          |
|-----|------------|------|--|---|---|--------------|--------------|---------------------------|--------|---|----------------------------------|
| 528 | cg43969763 | 2663 | CAAAGCAAGAA<br>GTTCTTGGAGAA[G/<br>T]TGCCAGATGGC<br>ACTGGAACTTAA        | G | T | Lys<br>(757) | Asn<br>(757) | NON-<br>CONSER-<br>VATIVE | kinase | Human Gene SWISSPROT-<br>ID:Q13627<br>SERINE/THREONINE-SPECIFIC<br>PROTEIN KINASE MINIBRAIN<br>HOMOLOG (EC 2.7.1.-) (HP86)<br>(DYRK) - HOMO SAPIENS<br>(HUMAN), 763 aa.           | 0<br>21<br>(21q22.1<br>)         |
| 529 | cg43932396 | 1226 | AGTCCACCGCCGC<br>CTCAGGGCGGTGQC<br>/TGGTGGCGAGT<br>AGGAGAACCTGGGG<br>G | C | T | Gly<br>(758) | Ser<br>(758) | NON-<br>CONSER-<br>VATIVE | kinase | Human Gene SWISSPROT-<br>ID:P31749 RAC-ALPHA<br>SERINE/THREONINE KINASE<br>(EC 2.7.1.-) (RAC-PK-ALPHA)<br>(PROTEIN KINASE B) (PKB) (C-<br>AKT) - HOMO SAPIENS<br>(HUMAN), 480 aa. | 1.40E-262<br>14<br>(14q32.3<br>) |
| 530 | cg43917871 | 1429 | GGCACTGAAGAAA<br>TCCCTGACATCA[T/<br>C]ATTGGCGCTGCT<br>GACGGGGCTACTG    | T | C | Met<br>(759) | Val<br>(759) | NON-<br>CONSER-<br>VATIVE | kinase | Human Gene SWISSPROT-<br>ID:P19138 CASEIN KINASE II,<br>ALPHA CHAIN (CK II) (EC<br>2.7.1.37) - HOMO SAPIENS<br>(HUMAN), AND BOS TAURUS<br>(BOVINE), 391 aa.                       | 2.00E-215<br>11<br>(20p13)       |
| 531 | cg43917871 | 1621 | GGGCTGACAAGGT<br>GCTGATTTCAC[T/<br>G]GTGGACAAAGC<br>GTTCCCCATCGCTT     | T | G | Ser<br>(760) | Arg<br>(760) | NON-<br>CONSER-<br>VATIVE | kinase | Human Gene SWISSPROT-<br>ID:P19138 CASEIN KINASE II,<br>ALPHA CHAIN (CK II) (EC<br>2.7.1.37) - HOMO SAPIENS<br>(HUMAN), AND BOS TAURUS<br>(BOVINE), 391 aa.                       | 2.00E-215<br>11<br>(20p13)       |

|     |            |      |  |   |   |     |              |                          |               |   |           |                     |
|-----|------------|------|--|---|---|-----|--------------|--------------------------|---------------|---|-----------|---------------------|
| 532 | cg43917871 | 1713 | TTC AAT GTGT TATT<br>TGT CAATA TAG [TC<br>[C] ATATAAAJCCTC<br>TGTCCCCAGAAC | T | C | Asp | Gly<br>(761) | NON-<br>CONSER-<br>ATIVE | kinase        | Human Gene SWISSPROT-<br>ID:P19138 CASEIN KINASE II,<br>ALPHA CHAIN (CK II) (EC<br>2.7.1.37) - HOMO SAPIENS<br>(HUMAN), AND BOS TAURUS<br>(BOVINE), 391 aa.   | 2.00E-215 | 11<br>(20p13)       |
| 533 | cg43917871 | 2096 | TGT AAAAATCGAAT<br>ATCATAGCTCTGT[V<br>C] AACGCTCTGGTAC<br>AATGCCCTGAAGT    | T | G | Leu | Phe<br>(762) | NON-<br>CONSER-<br>ATIVE | kinase        | Human Gene SWISSPROT-<br>ID:P19138 CASEIN KINASE II,<br>ALPHA CHAIN (CK II) (EC<br>2.7.1.37) - HOMO SAPIENS<br>(HUMAN), AND BOS TAURUS<br>(BOVINE), 391 aa.   | 2.00E-215 | 11<br>(20p13)       |
| 534 | cg43322545 | 1107 | TGG GCT AGGCAGC<br>CTCCATCC[C]A/<br>C]CCCCTTATCACCA<br>TCCGCGTGCGATG       | A | C | Thr | Pro<br>(763) | NON-<br>CONSER-<br>ATIVE | kinaserceptor | Human Gene SWISSPROT-<br>ID:P30530 TYROSINE-PROTEIN<br>KINASE RECEPTOR UFO<br>PRECURSOR (EC 2.7.1.112)<br>(AXL ONCOGENE) - HOMO<br>SAPIENS (HUMAN), 887<br>aa. pcds:SWISSPROT-ID:P30530<br>TYROSINE-PROTEIN KINASE<br>RECEPTOR UFO PRECURSOR<br>(EC 2.7.1.112) (AXL<br>ONCOGENE) - HOMO SAPIENS<br>(HUMAN), 887 aa. | 0         | 19<br>(19q13.1<br>) |

|     |            |      |   |   |   |     |              |                           |                |   |   |                     |
|-----|------------|------|---|---|---|-----|--------------|---------------------------|----------------|---|---|---------------------|
| 535 | cg43322545 | 2116 | TTCCTCCATTIC<br>CCGGCTCGGGIA/<br>GCCCAGCCAGTGA<br>CCTGCCCACTCAG | A | G | Asp | Gly<br>(764) | NON-<br>CONSER-<br>VATIVE | kinasereceptor | Human Gene SWISSNEW-<br>ID:P30530 TYROSINE-PROTEIN<br>KINASE RECEPTOR UFO<br>PRECURSOR (EC 2.7.1.112)<br>(AXL ONCOGENE) - HOMO<br>SAPIENS (HUMAN), 887<br>aa pcls:SWISSPROT;ID:P30530<br>TYROSINE-PROTEIN KINASE<br>RECEPTOR UFO PRECURSOR<br>(EC 2.7.1.112) (AXL<br>ONCOGENE) - HOMO SAPIENS<br>(HUMAN), 887 aa. | 0 | 19<br>(19q13.1<br>) |
|     |            |      |   |   |   |     |              |                           |                |   |   |                     |
|     |            |      |   |   |   |     |              |                           |                |   |   |                     |
|     |            |      |   |   |   |     |              |                           |                |   |   |                     |
|     |            |      |   |   |   |     |              |                           |                |   |   |                     |

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|-----|------------|-----|---|------------------|---------------------|---------------------------|---------|---|-----------|---------------|
| 536 | cg43958558 | 863 | TCCGGGATAAGCT<br>CAGGTGCTCCAIG<br>TGGTAGGGCCT<br>GAGGTGCTGTC<br>C     | G<br>T<br>T<br>G | Pro<br>His<br>(765) | NON-<br>CONSER-<br>VATIVE | laminin | Human Gene Homologous to<br>SWISSPROT-ID:P17931<br>GALECTIN-3 (GALACTOSE-<br>SPECIFIC LECTIN 3) (MAC-2<br>ANTIGEN) (IGE-BINDING<br>PROTEIN) (35 KD LECTIN)<br>(CARBOHYDRATE BINDING<br>PROTEIN 35) (CBP 35)<br>(LAMININ-BINDING PROTEIN)<br>(LECTIN L-29) (L-31)<br>(GALACTOSIDE-BINDING<br>PROTEIN) (GALBP) - HOMO<br>SAPIENS (HUMAN), 249<br>aa-[pels:SWISSPROT-ID:P17931<br>GALECTIN-3 (GALACTOSE-<br>SPECIFIC LECTIN 3) (MAC-2<br>ANTIGEN) (IGE-BINDING<br>PROTEIN) (35 KD LECTIN)<br>(CARBOHYDRATE BINDING<br>PROTEIN 35) (CBP 35)<br>(LAMININ-BINDING PROTEIN)<br>(LECTIN L-29) (L-31)<br>(GALACTOSIDE-BINDING<br>PROTEIN) (GALBP) - HOMO<br>SAPIENS (HUMAN), 249 aa. | 3.90E-139 | 14<br>(14q21) |
| 537 | cg43966144 | 718 | AAGCTTGATGCC<br>CTCACAGCAGTGC<br>AAGCACAAGACTG<br>CCCAGCCCCAATGG<br>A | C<br>A<br>A<br>A | Glu<br>(766)        | NON-<br>CONSER-<br>VATIVE | MHC     | Human Gene Homologous to<br>SWISSPROT-ID:P28068 CLASS<br>II HISTOCOMPATIBILITY<br>ANTIGEN M BETA CHAIN<br>PRECURSOR - HOMO SAPIENS<br>(HUMAN), 263 aa.  | 9.10E-147 | 6<br>(6p21.3) |

|     |            |      |   |   |   |     |              |                           |              |  |           |               |
|-----|------------|------|---|---|---|-----|--------------|---------------------------|--------------|--|-----------|---------------|
| 538 | cg43966144 | 823  | ACTTACACCTGTGT<br>GGTAGAGCACAT/<br>CTGGGGCTCCCTGA<br>GCCCATCCCTCGG      | T | C | Ile | Thr<br>(767) | NON-<br>CONSER-<br>VATIVE | MHC          | Human Gene Homologous to<br>SWISSPROT-ID:P28068 CLASS<br>II HISTOCOMPATIBILITY<br>ANTIGEN, M BETA CHAIN<br>PRECURSOR - HOMO SAPIENS<br>(HUMAN), 263 aa.                            | 9.10E-147 | 6<br>(6p21.3) |
| 539 | cg42686658 | 907  | GGCCTGGGGCT<br>TCCTCGGGCA[C/<br>T]CGTCCTCAT<br>CATGGGCACATAT            | C | T | Thr | Ile<br>(768) | NON-<br>CONSER-<br>VATIVE | MHC          | Human Gene Homologous to<br>SWISSPROT-ID:P06340 HLA<br>CLASS II<br>HISTOCOMPATIBILITY<br>ANTIGEN, DZ ALPHA CHAIN<br>PRECURSOR (MHC DN-ALPHA)<br>- HOMO SAPIENS (HUMAN),<br>250 aa. | 3.70E-134 | 6<br>(6p21.3) |
| 540 | cg38337333 | 1044 | CTGAGCCCCAGAGC<br>GTTGCTCCTGC/C/<br>GTCATGAGCACCAC<br>AGTCAGGGCTTG      | C | G | Pro | Ala<br>(769) | NON-<br>CONSER-<br>VATIVE | MHC          | Human Gene Homologous to<br>SPTREMBL-ID:Q95368 HLA<br>CLASS I INHIBITORY NK<br>RECEPTOR - HOMO SAPIENS<br>(HUMAN), 455 aa.   | 1.80E-113 | 19            |
| 541 | cg38337333 | 424  | AGCCCCGGCGGGC<br>CCCACGGTTCGC[A/<br>G]CAGGGAGAGAAC<br>GTGACCTTGTCCTG    | A | G | Thr | Ala<br>(770) | NON-<br>CONSER-<br>VATIVE | MHC          | Human Gene Homologous to<br>SPTREMBL-ID:Q95368 HLA<br>CLASS I INHIBITORY NK<br>RECEPTOR - HOMO SAPIENS<br>(HUMAN), 455 aa.   | 1.80E-113 | 19            |
| 542 | cg42481172 | 340  | CCGGCTGTGCTCA<br>GGGGTGTGGGTIA<br>(G)CGGATAACAGAG<br>GAGGGGCTGGTGG<br>A | A | G | Thr | Ala<br>(771) | NON-<br>CONSER-<br>VATIVE | misc_channel | Human Gene Similar to<br>SPTREMBL-ID:P91197 SIMILAR<br>TO LIGAND-GATED IONIC<br>CHANNEL PROTEIN -<br>CAENORHABDITIS ELEGANS,<br>461 aa.  | 2.30E-71  | 1             |

|     |            |      |   |   |   |     |              |                           |              |  |           |                     |
|-----|------------|------|---|---|---|-----|--------------|---------------------------|--------------|--|-----------|---------------------|
| 543 | cg3000465  | 238  | GAAGATGCCCTCC<br>TCAGACATGAGTGC<br>TGAAAGGTATC<br>AGAAATGGTCCG<br>C     | G | T | Trp | Leu<br>(772) | NON-<br>CONSER-<br>VATIVE | misc_channel | Human Gene Similar to<br>SPTREMBL-ID:P91197 SIMILAR<br>TO LIGAND-GATED IONIC<br>CHANNEL PROTEIN -<br>CAENORHABDITIS ELEGANS,<br>461 aa.  | 6.10E-70  | 8<br>(8p11.2)       |
| 544 | cg3000465  | 240  | AGATGCCCTCC<br>AGACATGAGTGGI<br>A/CIAAGGTATCA<br>GAAATGGTCCGC<br>CC     | A | C | Lys | Gln<br>(773) | NON-<br>CONSER-<br>VATIVE | misc_channel | Human Gene Similar to<br>SPTREMBL-ID:P91197 SIMILAR<br>TO LIGAND-GATED IONIC<br>CHANNEL PROTEIN -<br>CAENORHABDITIS ELEGANS,<br>461 aa.  | 6.10E-70  | 8<br>(8p11.2)       |
| 545 | cg43249083 | 1067 | GCCCTGGGCTCC<br>ACTACGGTTGCTA<br>TTCGCGTGGAGG<br>GCTGCAAGGGCT<br>T      | A | T | His | Leu<br>(774) | NON-<br>CONSER-<br>VATIVE | nucl_recept  | Human Gene SWISSPROT-<br>ID:P20393 V-ERBA RELATED<br>PROTEIN EAR-1 -HOMO<br>SAPIENS (HUMAN), 614 aa.   | 0         | 17<br>(17q11.2<br>) |
| 546 | cg44928796 | 68   | AGCGGGACGGTCC<br>GGAGCAAGCCCCA<br>G/CJAGGCAGAGGA<br>GGCGACAGAGGGA<br>AA | G | C | Gln | His<br>(775) | NON-<br>CONSER-<br>VATIVE | nucl_recept  | Human Gene SWISSNEW-<br>ID:P10275 ANDROGEN<br>RECEPTOR - HOMO SAPIENS<br>(HUMAN), 919<br>aa. pcis:SWISSPROT-ID:P10275<br>ANDROGEN RECEPTOR -<br>HOMO SAPIENS (HUMAN), 919<br>aa. | 0         | X (Xq11)            |
| 547 | cg43323772 | 91   | GTCGGGGAGTGA<br>GCGATGAGCTGGC<br>TTTCTGTTCTGG<br>CCCACAGTCGC            | C | T | Leu | Phe<br>(776) | NON-<br>CONSER-<br>VATIVE | nuclease     | Human Gene TREMBLNEW-<br>ID:G2935442 RIBONUCLEASE<br>H1 - HOMO SAPIENS (HUMAN),<br>286 aa. pcis:TREMBL NEW-<br>ID:G2935444 RIBONUCLEASE<br>H1 - HOMO SAPIENS (HUMAN),<br>286 aa. | 1.40E-157 |                     |

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|-----|------------|-----|---|---|---|--------------|--------------|---------------------------|------------|---|-----------|---------------------|
| 548 | cg42732993 | 809 | GGCTATAATCACA<br>ATGGGGAAATGGTG<br>TTGAAAGCCAAA<br>CCAAAAATGGCCA<br>A | G | T | Cys<br>(77)  | Phe<br>(77)  | NON-<br>CONSER-<br>VATIVE | oncogene   | Human Gene Homologous to<br>SPTREMBL-ID:Q13692<br>BCR/ABL FUSION PROTEIN -<br>HOMO SAPIENS (HUMAN), 284<br>aa (fragment).   | 6.00E-150 |                     |
| 549 | cg42904626 | 155 | ATATAAACCTGTG<br>GTAGTTGGAGCT[G<br>T]GTGGCGTAGGC<br>AAGAGTGCTTGA<br>C | G | T | Gly<br>(77)  | Cys<br>(77)  | NON-<br>CONSER-<br>VATIVE | oncogene   | Human Gene Similar to<br>SWISSPROT-ID:P01118<br>TRANSFORMING PROTEIN<br>P21/K-RAS 2B - HOMO<br>SAPIENS (HUMAN), 188 aa.   | 1.10E-97  | 12                  |
| 550 | cg42904626 | 304 | TGGATAATTCCTGGAC<br>ACAGGAGGTCA[A/<br>C]GAGGAGTACAGT<br>GCAATGAGGGACC | A | C | Gln<br>(77)  | His<br>(77)  | NON-<br>CONSER-<br>VATIVE | oncogene   | Human Gene Similar to<br>SWISSPROT-ID:P01118<br>TRANSFORMING PROTEIN<br>P21/K-RAS 2B - HOMO<br>SAPIENS (HUMAN), 188 aa.   | 1.10E-97  | 12                  |
| 551 | cg42691989 | 706 | CTGTTCAGGATCTC<br>CTCATTCTGACAT/T<br>GGTTCTCCTGATGT<br>CCAAATTGTTG    | A | T | Cys<br>(780) | Ser<br>(780) | NON-<br>CONSER-<br>VATIVE | peroxidase | Human Gene Homologous to<br>SWISSPROT-ID:P18283<br>GLUTATHIONE PEROXIDASE-<br>GASTROINTESTINAL (EC<br>1.11.1.9) (GSHPX-GI)<br>(GLUTATHIONE PEROXIDASE-<br>RELATED PROTEIN 2) (GPRP) -<br>HOMO SAPIENS (HUMAN), 190<br>aa. | 8.90E-101 | 14<br>(14q24.1<br>) |

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| 552 | cg43917453 | 4096 | AGGTCCCTCGCGGA<br>GCTGGGGTCTCGGGIA<br>GICCCGGGGAGGGT<br>AGGTCAAGCGCAGA<br>C | A | G | Ser<br>(781) | Pro<br>(781) | NON-<br>CONSER-<br>VATIVE | phosphatase   | Human Gene TREMBL NEW-<br>ID:G2262075 IAR/RECEPTOR-<br>LIKE PROTEIN-TYROSINE<br>PHOSPHATASE PRECURSOR -<br>HOMO SAPIENS (HUMAN),<br>1015 aa.  | 0         | 7             |
| 553 | cg43947363 | 368  | CTGGCGCACTACT<br>CGGACCTGCTCC[C/<br>T]CCRGCGGGCCT<br>GGGGCTGATTGAG          | C | T | Gly<br>(782) | Glu<br>(782) | NON-<br>CONSER-<br>VATIVE | phosphatase   | Human Gene SWISSPROT-<br>ID:P23469 PROTEIN-TYROSINE<br>PHOSPHATASE EPSILON<br>PRECURSOR (EC 3.1.3.48) (R-<br>PTP- EPSILON) - HOMO<br>SAPIENS (HUMAN), 700 aa.   | 0         |               |
| 554 | cg43928335 | 3187 | GCACAAGGAACGG<br>AATTGCTGTCTGT[A/<br>G]TTCTGCTTTAA<br>CAGGATTGATGTC         | A | G | Ile<br>(783) | Thr<br>(783) | NON-<br>CONSER-<br>VATIVE | phosphatase   | Human Gene SWISSPROT-<br>ID:P54613 PROTEIN<br>PHOSPHATASE PP2A, 65 KD<br>REGULATORY SUBUNIT,<br>BETA ISOFORM (PROTEIN<br>PHOSPHATASE PP2A SUBUNIT<br>A, BETA ISOFORM) (P65-BETA)<br>- SUS SCROFA (PIG), 602 aa<br>(fragment). | 3.20E-302 | 11<br>(11q22) |
| 555 | cg43996195 | 1330 | CTTCGGGGAAAGT<br>TGGGGATTCAAC[C/<br>T]GTAGTCAAAGAT<br>CTGGGCCCTGAGTT        | C | T | Gly<br>(784) | Ser<br>(784) | NON-<br>CONSER-<br>VATIVE | phosphorylase | Human Gene SWISSPROT-<br>ID:P00491 PURINE<br>NUCLEOSIDE<br>PHOSPHORYLASE (EC 2.4.2.1)<br>(INOSINE PHOSPHORYLASE)<br>(PNP) - HOMO SAPIENS<br>(HUMAN), 289 aa.  | 2.40E-155 |               |

|     |            |      |   |   |   |     |              |                           |            |  |   |                     |
|-----|------------|------|---|---|---|-----|--------------|---------------------------|------------|--|---|---------------------|
| 556 | cg44022214 | 3340 | AGGTCCCTCTCGA<br>ATTGGGATGCCJA<br>/GAGGTGCATCAT<br>CATCATCCCAGAG<br>G | A | G | Trp | Arg<br>(785) | NON-<br>CONSER-<br>VATIVE | polymerase | Human Gene SWISSNEW-<br>ID:P28340 DNA POLYMERASE<br>DELTA CATALYTIC CHAIN<br>(EC 2.7.7.7) - HOMO SAPIENS<br>(HUMAN), 1107<br>aa.[p]c;s:SWISSPROT;ID:P28340<br>DNA POLYMERASE DELTA<br>CATALYTIC CHAIN (EC 2.7.7.7)<br>- HOMO SAPIENS (HUMAN),<br>1107 aa.  | 0 | 19<br>(19q13.3<br>) |
| 557 | cg43958858 | 1593 | CTCAGACCATGTC<br>CTTCGGATGCAC[C/<br>G]GTTACAGAGCAC<br>CTGGGGAGCAGGA   | C | G | Arg | Gly<br>(786) | NON-<br>CONSER-<br>VATIVE | polymerase | Human Gene SWISSNEW-<br>ID:P25205 DNA REPLICATION<br>LICENSING FACTOR MCM3<br>(DNA POLYMERASE ALPHA<br>HOLOENZYME-ASSOCIATED<br>PROTEIN1) (RLF BETA<br>SUBUNIT) (P102 PROTEIN) -<br>HOMO SAPIENS (HUMAN), 808<br>aa.[p]c;s:SWISSPROT;ID:P25205<br>DNA REPLICATION<br>LICENSING FACTOR MCM3<br>(DNA POLYMERASE ALPHA<br>HOLOENZYME-ASSOCIATED<br>PROTEIN1) (RLF BETA<br>SUBUNIT) (P102 PROTEIN) -<br>HOMO SAPIENS (HUMAN), 808<br>aa. | 0 | 6 (6p12)            |
|     |            |      |   |   |   |     |              |                           |            |  |   |                     |
|     |            |      |   |   |   |     |              |                           |            |  |   |                     |
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|-----|------------|-----|---|---|---|-----|--------------|---------------------------|------------------------|---|----------|---------------|
| 558 | cg42534568 | 641 | CGCTTITGAAAGCC<br>AGCTGGGCACCCIA<br>TTGGCGCAGTTCCC<br>CAACACACTCCTG     | A | T | Gln | Leu<br>(787) | NON-<br>CONSER-<br>VATIVE | potassium Chan-<br>nel | Human Gene SWISSPROT-<br>ID:P22460 VOLTAGE-GATED<br>POTASSIUM CHANNEL<br>PROTEIN KV1.5 (HK2) (HPCN1)<br>- HOMO SAPIENS (HUMAN),<br>613 aa.  | 0        | 12<br>(12p13) |
| 559 | cg42534568 | 868 | GGGGGACGAGGCC<br>ATGGGAGCCTTC[C<br>/G]GGCAGGATGAG<br>GGCTTCATAAAG<br>A  | C | G | Arg | Gly<br>(788) | NON-<br>CONSER-<br>VATIVE | potassium Chan-<br>nel | Human Gene SWISSPROT-<br>ID:P22460 VOLTAGE-GATED<br>POTASSIUM CHANNEL<br>PROTEIN KV1.5 (HK2) (HPCN1)<br>- HOMO SAPIENS (HUMAN),<br>613 aa.  | 0        | 12<br>(12p13) |
| 560 | cg42534568 | 910 | CATTAAAGAAAGAG<br>GAGAAGGCCCTGIC<br>/GICCCCGCAACGAG<br>TTCAGGCCAGG<br>T | C | G | Pro | Ala<br>(789) | NON-<br>CONSER-<br>VATIVE | potassium Chan-<br>nel | Human Gene SWISSPROT-<br>ID:P22460 VOLTAGE-GATED<br>POTASSIUM CHANNEL<br>PROTEIN KV1.5 (HK2) (HPCN1)<br>- HOMO SAPIENS (HUMAN),<br>613 aa.  | 0        | 12<br>(12p13) |
| 561 | cg43154190 | 898 | TGGAGGGGATGCT<br>CATTTTGATGAA[G/<br>C]ATGAAAGGTGG<br>ACCAACAATTCA<br>G  | G | C | Asp | His<br>(790) | NON-<br>CONSER-<br>VATIVE | protease               | Human Gene Similar to<br>SWISSPROT-ID:P50280<br>MATRILYSIN PRECURSOR (EC<br>3.4.24.23) (PUMP-1 PROTEASE)<br>(UTERINE<br>METALLOPROTEINASE)<br>(MATRIX<br>METALLOPROTEINASE-7)<br>(MMP-7) (MATRIN) - RATTUS<br>NORVEGICUS (RAT), 267 aa. | 2.40E-59 | 11<br>(11q22) |

|     |            |      |   |   |   |              |              |                  |           |  |           |               |
|-----|------------|------|---|---|---|--------------|--------------|------------------|-----------|--|-----------|---------------|
| 562 | cg43154190 | 923  | GATGAAAGGTGGACCAACAATTCA[GC]AGTACATCGTGTGCG                   | G | C | Arg<br>(791) | Thr<br>(791) | NON-CONSERVATIVE | protease  | Human Gene Similar to SWISSPROT-ID:P50280 MATRIX YSIN PRECURSOR (EC 3.4.24.23) (PUMP-1 PROTEASE) (UTERINE MATRIX METALLOPROTEINASE-7) (MMP-7) (MATRIN) - RAT/TUS NORVEGICUS (RAT), 267 aa.                                     | 2.40E-59  | 11<br>(11q22) |
| 563 | cg43927549 | 694  | ATTCTACGATTCCGTTGCTCCAG[G/T]GTAACCTAGCGCTCCTTCCGTAAC          | G | T | Gly<br>(792) | Cys<br>(792) | NON-CONSERVATIVE | reductase | Human Gene Homologous to SWISSPROT-ID:P16083 NAD(PH) DEHYDROGENASE (QUINONE) 2 (EC 1.6.9.2) (QUINONE REDUCTASE) (DT-DIAPHORASE) (AZOREDUCTASE) (PHYLLOQUINONE REDUCTASE) (MENADIONE REDUCTASE) - HOMO SAPIENS (HUMAN), 231 aa. | 1.60E-124 | 6 (6pter)     |
| 564 | cg43325541 | 1081 | CGCTTGCCITTCCTCCC[GAAAGGTCTGCC[T]CCTTCACGGCTT]CGGCTTCCCCGCAAG | C | T | Gly<br>(793) | Glu<br>(793) | NON-CONSERVATIVE | synthase  | Human Gene TREMBL NEW-ID:G2725625 ACETOLACTATE SYNTHASE - HOMO SAPIENS (HUMAN), 632 aa.  | 0         | 19            |

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| 565 | cg43064068 | 1474 | GTGAAGGCATTG<br>TGGTCCCTGGCCTTC/<br>TGCAAGTCCCTGTC<br>CCATGACCCAGAA        | C | T | Ser | Leu<br>(794) | NON-<br>CONSER-<br>VATIVE | synthase | Human Gene Similar to<br>SWISSNEW-ID:P39062<br>ACETYL-COENZYME A<br>SYNTHETASE (EC 6.2.1.1)<br>(ACETATE--COA LIGASE)<br>(ACYL- ACTIVATING<br>ENZYME) (ACETYL-COA<br>SYNTHASE) - BACILLUS<br>SUBTILIS, 572<br>aa. pcis:SWISSPROT-ID:P39062 | 7.40E-65 |
| 566 | cg43064068 | 1617 | GACTGTACACGGG<br>AAAATCCAACGAIG<br>/A/C/C/AAGCTTCGA<br>GACAAGGAGTGGAA<br>A | G | A | Ala | Thr<br>(795) | NON-<br>CONSER-<br>VATIVE | synthase | Human Gene Similar to<br>SWISSNEW-ID:P39062<br>ACETYL-COENZYME A<br>SYNTHETASE (EC 6.2.1.1)<br>(ACETATE--COA LIGASE)<br>(ACYL- ACTIVATING<br>ENZYME) (ACETYL-COA<br>SYNTHASE) - BACILLUS<br>SUBTILIS, 572<br>aa. pcis:SWISSPROT-ID:P39062 | 7.40E-65 |

|     |            |      |   |   |   |              |              |                          |     |   |           |               |
|-----|------------|------|---|---|---|--------------|--------------|--------------------------|-----|---|-----------|---------------|
| 567 | cg36988276 | 1119 | GCAAGAAACTGAT<br>TATATGACTCAGA<br>/GCTTAGGGTCAG<br>AGATCCTCTGGC       | A | G | Thr<br>(796) | Ala<br>(796) | NON-<br>CONSER-<br>ATIVE | tm7 | Human Gene SWISSPROT-<br>ID:P23945 FOLLICLE<br>STIMULATING HORMONE<br>RECEPTOR PRECURSOR (FSH-<br>R) (FOLLITROPIN RECEPTOR)-<br>HOMO SAPIENS (HUMAN), 695<br>aa.      | 0         | 2 (2p21)      |
| 568 | cg36988276 | 535  | AAGGCCAACAAACC<br>TGCTCTACATCA[A/<br>C]CCCTGAGGCCTT<br>CCAGAACCTCCC   | A | C | Asn<br>(797) | Thr<br>(797) | NON-<br>CONSER-<br>ATIVE | tm7 | Human Gene SWISSPROT-<br>ID:P23945 FOLLICLE<br>STIMULATING HORMONE<br>RECEPTOR PRECURSOR (FSH-<br>R) (FOLLITROPIN RECEPTOR)-<br>HOMO SAPIENS (HUMAN), 695<br>aa.      | 0         | 2 (2p21)      |
| 569 | cg32296848 | 1475 | GAATGTCTTGAGA<br>ATCCAGTGTCTC[C/<br>T]GCAGAAAGCAGT<br>CTTCCAAACATGC   | C | T | Arg<br>(798) | Cys<br>(798) | NON-<br>CONSER-<br>ATIVE | tm7 | Human Gene SWISSPROT-<br>ID:P35348 ALPHA-1A<br>ADRENERGIC RECEPTOR<br>(ALPHA 1A-ADRENOCEPTOR)<br>(ALPHA-1C ADRENERGIC<br>RECEPTOR) - HOMO SAPIENS<br>(HUMAN), 466 aa. | 1.60E-252 | 8 (8p21)      |
| 570 | cg2524739  | 1590 | TCTCTCTGGAGAA<br>GATCCAACCCATC<br>/GACACAAACCGG<br>TCAGGCACCAACC<br>T | C | G | Ile<br>(799) | Met<br>(799) | NON-<br>CONSER-<br>ATIVE | tm7 | Human Gene SWISSPROT-<br>ID:P21728 D(1A) DOPAMINE<br>RECEPTOR - HOMO SAPIENS<br>(HUMAN), 446 aa.  | 8.30E-240 | 5<br>(5q35.1) |

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|-----|------------|-----|---|---|---|--------------|--------------|---------------------------|-----|---|-----------|
| 571 | cg2320320  | 394 | AGTGTCTGGATGA<br>TCTTTGTGTCAC/C/<br>TGTGCATCCGGTT<br>CACAAATGGCCTT    | C | T | Thr<br>(800) | Ile<br>(800) | NON-<br>CONSER-<br>VATIVE | tm7 | Human Gene SWISSPROT-<br>ID: P04001 GREEN-SENSITIVE<br>OPSIN (GREEN CONE<br>PHOTORECEPTOR PIGMENT)-<br>HOMO SAPIENS (HUMAN), 364<br>aa.   | 8.50E-199 |
| 572 | cg43264978 | 519 | CATCTTCCTCATCA<br>ACCTCTTCAAGCAG<br>GCATTTCCTCCTC<br>ACGTGGATGAG      | A | G | Ser<br>(801) | Gly<br>(801) | NON-<br>CONSER-<br>VATIVE | tm7 | Human Gene TREMBL NEW-<br>ID: G2736282 G PROTEIN<br>COUPLED RECEPTOR - HOMO<br>SAPIENS (HUMAN), 362 aa.   | 1.40E-196 |
| 573 | cg3003708  | 285 | TCTTTTGCGACAT<br>CTGCTCTCTCT[C]<br>CAACCACCGTCCCC<br>AAGATGCTGGCC     | T | C | Phe          | Ser<br>(802) | NON-<br>CONSER-<br>VATIVE | tm7 | Human Gene TREMBL NEW-<br>ID: E1246031 OLFACTORY<br>RECEPTOR - HOMO SAPIENS<br>(HUMAN), 312 aa.   | 2.50E-160 |
| 574 | cg38841806 | 68  | GGCCCTGAGGCA<br>ACACCAACGGGCAT<br>/CICACAGCCTCTC<br>CATGCCAGCTGG      | T | C | Ile          | Thr<br>(803) | NON-<br>CONSER-<br>VATIVE | tm7 | Human Gene Similar to<br>SWISSPROT-ID: P30975<br>TACHYKININ-LIKE PEPTIDES<br>RECEPTOR 99D (DTKR) -<br>DROSOPHILA<br>MELANOGLASTER (FRUIT FLY),<br>519 aa.                       | 2.10E-67  |
| 575 | cg43336100 | 688 | TGGAAAGCGTGCAT<br>CCAGTGAGACCAQA<br>/TGTAGGGCTTGAGT<br>CTTITAGTGCCCTG | A | T | Met          | Leu<br>(804) | NON-<br>CONSER-<br>VATIVE | tnf | Human Gene SWISSPROT-<br>ID: P26022 PENTAXIN-<br>RELATED PROTEIN PTX3<br>PRECURSOR (TUMOR<br>NECROSIS FACTOR-<br>INDUCIBLE PROTEIN TSG-14)-<br>HOMO SAPIENS (HUMAN), 381<br>aa. | 2.20E-207 |
|     |            |     |   |   |   |              |              |                           |     |   | 3 (3q25)  |

|     |            |      |   |   |              |              |                          |                          |   |           |                     |
|-----|------------|------|---|---|--------------|--------------|--------------------------|--------------------------|---|-----------|---------------------|
| 576 | cg43335562 | 234  | GAGGGCGGGGAG T<br>CCAGGCCCTGGCCT<br>CJCCGGGTCCCCA<br>AGACCCTTGGCTC    | C | Leu<br>(805) | Pro<br>(805) | NON-<br>CONSER-<br>ATIVE | tnfceptor                | Human Gene Similar to<br>TREMBl, NEW-ID:G2653845 TNF<br>RECEPTOR-RELATED<br>RECEPTOR FOR TRAIL - HOMO<br>SAPIENS (HUMAN), 386 aa.   | 2.30E-55  | 8                   |
| 577 | cg43140548 | 2857 | ACTCGCACGTGGA<br>TCCTGAGGTGT[A/<br>G]AGAGTAAGGA<br>AGGCCTTGCCACA<br>G | A | G            | Tyr<br>(806) | His<br>(806)             | transcriptfactor         | Human Gene SPTREMBL-<br>ID:Q14872 METAL-<br>REGULATORY<br>TRANSCRIPTION FACTOR -<br>HOMO SAPIENS (HUMAN), 753<br>aa.  | 0         | 1                   |
| 578 | cg43011561 | 1285 | CATTGACAGCQAG<br>GCCTCCCTAGCC[C/<br>T]TCTTCATGGCGA<br>AGAAGAAAGACGCC  | C | T            | Leu<br>(807) | Phe<br>(807)             | NON-<br>CONSER-<br>ATIVE | Human Gene SWISSPROT-<br>ID:P35269 TRANSCRIPTION<br>INITIATION FACTOR IIF,<br>ALPHA SUBUNIT (IFIIF-<br>ALPHA) (TRANSCRIPTION<br>INITIATION FACTOR RAP74)-<br>HOMO SAPIENS (HUMAN), 517<br>aa. | 4.30E-275 | 19<br>(19p13.3<br>) |
| 579 | cg43998970 | 1346 | TGACAGAGCTGTA<br>CCGTGACATTTTC/<br>GICAGGCACCTTCGG<br>GATGAATCAGGCA   | C | G            | Phe<br>(808) | Leu<br>(808)             | NON-<br>CONSER-<br>ATIVE | Human Gene SPTREMBL-<br>ID:Q07279 TRANSCRIPTION<br>FACTOR NF-E2 - MUS<br>MUSCULUS (MOUSE), 373 aa.  | 1.70E-177 | 12                  |

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| 580 | cg2537639 | 464 | CACTACTATGTCCTT<br>CACCGACCAAGC/C/<br>TGGCCGGGGTGCC<br>CCGGCTGACGGCTG | C | T | Pro<br>(809) | Leu<br>(809) | NON-<br>CONSER-<br>VATIVE | transferase | Human Gene SWISSPROT-<br>ID:P16442<br>FUCOSYLYCOPROTEIN<br>ALPHA-N-<br>ACETYLGLACTOSAMINYLT<br>RANSFERASE (EC 2.4.1.40)<br>(HISTO-BLOOD GROUP A<br>TRANSFERASE) (A<br>TRANSFERASE) /<br>FUCOSYLYCOPROTEIN 3-<br>ALPHA-<br>GALACTOSYLTRANSFERASE<br>(EC 2.4.1.37) (HISTO-BLOOD<br>GROUP B TRANSFERASE) (B<br>TRANSFERASE) (NAGAT)-<br>HOMO SAPIENS (HUMAN), 354<br>aa. | 6.50E-192 | 9 (9q34) |
|     |           |     |   |   |   |              |              |                           |             |   |           |          |
|     |           |     |   |   |   |              |              |                           |             |   |           |          |
|     |           |     |   |   |   |              |              |                           |             |   |           |          |
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|-----|-----------|-----|---|---|---|-----|--------------|---------------------------|-------------|--|-----------|----------|
| 581 | cg2537639 | 523 | TCGGCAGCTGTCA<br>GTGCTGGAGGTGIC<br>/GIGCGCCTACAAG<br>CGCTGGCAGGACG<br>T | C | G | Arg | Gly<br>(810) | NON-<br>CONSER-<br>VATIVE | transferase | Human Gene SWISSPROT-<br>ID:P16442<br>FUCOSYLGLYCOPROTEIN<br>ALPHA-N-<br>ACETYLGLACTOSAMINYLT<br>RANSFERASE (EC 2.4.1.40)<br>(HISTO-BLOOD GROUP A<br>TRANSFERASE) (A<br>TRANSFERASE) /<br>FUCOSYLGLYCOPROTEIN 3-<br>ALPHA-<br>GALACTOSYLTRANSFERASE<br>(EC 2.4.1.37) (HISTO-BLOOD<br>GROUP B TRANSFERASE) (B<br>TRANSFERASE) (NAGAT) -<br>HOMO SAPIENS (HUMAN), 354<br>aa. | 6.50E-192 | 9 (9q34) |
|-----|-----------|-----|---|---|---|-----|--------------|---------------------------|-------------|--|-----------|----------|

|     |           |     |  |   |   |     |              |                           |             |  |           |          |
|-----|-----------|-----|--|---|---|-----|--------------|---------------------------|-------------|--|-----------|----------|
| 582 | cg2537639 | 643 | GGTGTGCGGGAC<br>GTGGACATGGAGT<br>/ATCCCGCAC<br>GTGGCGTGGAGA<br>T | T | A | Phe | Ile<br>(811) | NON-<br>CONSER-<br>VATIVE | transferase | Human Gene SWISSPROT-<br>ID:P16442<br>FUCOSYGLYCOPROTEIN<br>ALPHA-N-<br>ACETYLGLACTOSAMINYLT<br>RANSFERASE (EC 2.4.1.40)<br>(HISTO-BLOOD GROUP A<br>TRANSFERASE) (A<br>TRANSFERASE)/<br>FUCOSYGLYCOPROTEIN 3-<br>ALPHA-<br>GALACTOSYLTRANSFERASE<br>(EC 2.4.1.37) (HISTO-BLOOD<br>GROUP B TRANSFERASE) (B<br>TRANSFERASE) (NAGAT)-<br>HOMO SAPIENS (HUMAN), 354<br>aa. | 6.50E-192 | 9 (9q34) |
|-----|-----------|-----|--|---|---|-----|--------------|---------------------------|-------------|--|-----------|----------|

|     |           |     |   |   |   |     |              |                           |             |  |           |          |
|-----|-----------|-----|---|---|---|-----|--------------|---------------------------|-------------|--|-----------|----------|
| 583 | cg2537639 | 700 | TCCGGCTGTCGGCA<br>CCCTGCACCCIG/<br>AGCTTCTACGGAA<br>GAGCCGGAGGC | G | A | Gly | Ser<br>(812) | NON-<br>CONSER-<br>VATIVE | transferase | Human Gene SWISSPROT-<br>ID:P16442<br>FUCOSYL GLYCOPROTEIN<br>ACETYL GALACTOSAMINYLT<br>TRANSFERASE (EC 2.4.1.40)<br>(HISTO-BLOOD GROUP A<br>TRANSFERASE) (A<br>TRANSFERASE) /<br>FUCOSYL GLYCOPROTEIN 3-<br>ALPHA-<br>GALACTOSYL TRANSFERASE<br>(EC 2.4.1.37) (HISTO-BLOOD<br>GROUP B TRANSFERASE) (B<br>TRANSFERASE) (NAGAT) -<br>HOMO SAPIENS (HUMAN), 354<br>aa. | 6.50E-192 | 9 (9q34) |
|-----|-----------|-----|---|---|---|-----|--------------|---------------------------|-------------|--|-----------|----------|

|     |           |     |  |   |   |     |              |                           |             |  |           |          |
|-----|-----------|-----|--|---|---|-----|--------------|---------------------------|-------------|--|-----------|----------|
| 584 | cg2537639 | 793 | CAAGGACCGAGGGC   C<br>GATTTCCTACTAC[C/<br>A]TGGGGGGGTCTCT<br>TCGGGGGGTCGGT | C | A | Leu | Met<br>(813) | NON-<br>CONSER-<br>VATIVE | transferase | Human Gene SWISSPROT-<br>ID:P16442<br>FUCOSYLYCOPROTEIN<br>ALPHA-N-<br>ACETYLGLACTOSAMINYLT<br>RANSFERASE (EC 2.4.1.40)<br>(HISTO-BLOOD GROUP A<br>TRANSFERASE) (A<br>TRANSFERASE) /<br>FUCOSYLYCOPROTEIN 3-<br>ALPHA-<br>GALACTOSYLTRANSFERASE<br>(EC 2.4.1.37) (HISTO-BLOOD<br>GROUP B TRANSFERASE) (B<br>TRANSFERASE) (NAGAT) -<br>HOMO SAPIENS (HUMAN), 354<br>aa. | 6.50E-192 | 9 (9q34) |
|     |           |     |  |   |   |     |              |                           |             |  |           |          |
|     |           |     |  |   |   |     |              |                           |             |  |           |          |
|     |           |     |  |   |   |     |              |                           |             |  |           |          |
|     |           |     |  |   |   |     |              |                           |             |  |           |          |

|     |            |      |  |   |   |              |              |                          |             |  |           |          |
|-----|------------|------|--|---|---|--------------|--------------|--------------------------|-------------|--|-----------|----------|
| 585 | cg2537639  | 826  | GTCCTCTGGGGG<br>TCGGGTGCAAGAGG<br>/ATGCAGGGCIC<br>ACCAAGGGCTGCC<br>A   | G | A | Val<br>(814) | Met<br>(814) | NON-<br>CONSER-<br>ATIVE | transferase | Human Gene SWISSPROT-<br>ID:P16442<br>FUCOSYLYCOPROTEIN<br>ALPHA-N-<br>ACETYLGLACTOSAMINYLT<br>RANSFERASE (EC 2.4.1.40)<br>(HISTO-BLOOD GROUP A<br>TRANSFERASE) (A<br>TRANSFERASE) /<br>FUCOSYLYCOPROTEIN 3-<br>ALPHA-<br>GALACTOSYLTRANSFERASE<br>(EC 2.4.1.37) (HISTO-BLOOD<br>GROUP B TRANSFERASE) (B<br>TRANSFERASE) (NAGAT)<br>HOMO SAPIENS (HUMAN), 354<br>aa. | 6.50E-192 | 9 (9q34) |
| 586 | cg42742340 | 3249 | CACATCGTTGG<br>AGCTGACCAGGC<br>/ATGACGGCTTGG<br>GCTCCAGGGGG<br>G       | C | A | Ala<br>(815) | Asp<br>(815) | NON-<br>CONSER-<br>ATIVE | transport   | Human Gene SWISSPROT-<br>ID:Q04671 P PROTEIN<br>(MELANOCYTE-SPECIFIC<br>TRANSPORTER PROTEIN)-<br>HOMO SAPIENS (HUMAN), 838<br>aa.  | 0         | 15       |
| 587 | cg41653463 | 427  | GTCTGAAGATT<br>CCACAAGGACATC<br>/G]CTGAAGCCCTC<br>ACCAAGGGAAAGAGC<br>C | C | G | Ile<br>(816) | Met<br>(816) | NON-<br>CONSER-<br>ATIVE | transport   | Human Gene SWISSPROT-<br>ID:P31641 SODIUM- AND<br>CHLORIDE-DEPENDENT<br>TAURINE TRANSPORTER-<br>HOMO SAPIENS (HUMAN), 620<br>aa.   | 0         | 3 (3p25) |

|     |            |      |   |   |   |              |              |                          |                   |  |   |               |
|-----|------------|------|---|---|---|--------------|--------------|--------------------------|-------------------|--|---|---------------|
| 588 | cg40351913 | 1165 | CAAGTTCACCAAC<br>AACTGCTACAGGIG<br>CTACGGCATTTGTC<br>ACCACCTCCATCA<br>A   | G | C | Asp<br>(817) | His<br>(817) | NON-<br>CONSER-<br>ATIVE | transport         | Human Gene SWISSPROT-<br>ID:Q01959 SODIUM-<br>DEPENDENT DOPAMINE<br>TRANSPORTER (DA<br>TRANSPORTER) (DAT) - HOMO<br>SAPIENS (HUMAN), 620 aa. | 0 | 5<br>(5p15.3) |
| 589 | cg40351913 | 1232 | TCCTCCGGTTCGT<br>CGCTCTCTCTCTT[C]<br>CCTGGGGTACATG<br>GCACAGAAAGCAC       | T | C | Phe<br>(818) | Ser<br>(818) | NON-<br>CONSER-<br>ATIVE | transport         | Human Gene SWISSPROT-<br>ID:Q01959 SODIUM-<br>DEPENDENT DOPAMINE<br>TRANSPORTER (DA<br>TRANSPORTER) (DAT) - HOMO<br>SAPIENS (HUMAN), 620 aa. | 0 | 5<br>(5p15.3) |
| 590 | cg43955093 | 4776 | CTGCCGGTAGCTGT<br>CCCAGGGCTCGGIC<br>/GICCGCGCCGCGCT<br>CGTCCATGTTGAG<br>G | C | G | Ala<br>(819) | Pro<br>(819) | NON-<br>CONSER-<br>ATIVE | UNCLASSIFI-<br>ED | Human Gene SWISSPROT-<br>ACC:Q16084 P130 - HOMO<br>SAPIENS (HUMAN), 1139 aa.   | 0 | 16            |
| 591 | cg43055918 | 522  | GCATAGGACATGG<br>CGGGCTTGGCCCTC/<br>CJCGCAGAGCTCTG<br>GGGGCTACTGCTA<br>G  | C | G | Gly<br>(820) | Arg<br>(820) | NON-<br>CONSER-<br>ATIVE | UNCLASSIFI-<br>ED | Human Gene SWISSPROT-<br>ACC:P42694 HYPOTHETICAL<br>PROTEIN KIAA0054 - Homo<br>sapiens (Human), 1942 aa.                                     | 0 | 17            |
| 592 | cg43968854 | 4604 | CAACCCCTAGAAC<br>ACCTGGCTGGCTT/<br>GJGAAAAGAGCTCTT<br>CCAGACACAGTA        | T | G | Leu<br>(821) | Ter<br>(821) | NON-<br>CONSER-<br>ATIVE | UNCLASSIFI-<br>ED | Human Gene SWISSNEW-<br>ACC:P46013 ANTIGEN KL-67 -<br>Homo sapiens (Human), 3256 aa.   | 0 | 10<br>(10q25) |

|     |            |      |  |   |   |              |              |                           |                   |  |   |          |
|-----|------------|------|--|---|---|--------------|--------------|---------------------------|-------------------|--|---|----------|
| 593 | cg43070241 | 1841 | CCATTGTTCAGAAGA<br>CATCCTTACGGTTTG<br>GGAATGCGCTGCA<br>AGCAAAATTGTCC   | T | G | Phe<br>(822) | Leu<br>(822) | NON-<br>CONSER-<br>VATIVE | UNCLASSIFI-<br>ED | Human Gene SWISSPROT-<br>ACC:P55157 MICROSMAL<br>TRIGLYCERIDE TRANSFER<br>PROTEIN, LARGE SUBUNIT<br>PRECURSOR - Homo sapiens<br>(Human), 894 aa. | 0 | 4 (4q22) |
| 594 | cg43262121 | 2001 | ACAATTCTAGAGAG<br>GGAGACTGAGCA<br>GTACACCCAGCAT<br>TGATCATGGTGC<br>AA  | G | T | Gln<br>(823) | His<br>(823) | NON-<br>CONSER-<br>VATIVE | UNCLASSIFI-<br>ED | Human Gene SPTREMBL-<br>ACC:Q15840 ZINC FINGER<br>PROTEIN BASONUCLIN -<br>HOMO SAPIENS (HUMAN), 994<br>aa.                                       | 0 |          |
| 595 | cg43262121 | 553  | ATCAGGAAAGGTG<br>TTGGATCACTGGTA<br>TGCGATCATGACC<br>AGTGAGGAAGAAG<br>T | A | T | Ser<br>(824) | Cys<br>(824) | NON-<br>CONSER-<br>VATIVE | UNCLASSIFI-<br>ED | Human Gene SPTREMBL-<br>ACC:Q15840 ZINC FINGER<br>PROTEIN BASONUCLIN -<br>HOMO SAPIENS (HUMAN), 994<br>aa.                                       | 0 |          |
| 596 | cg43262121 | 937  | CCCCAACAGGAA<br>GTCCATGGGCCJA<br>TGACCCCTGACAGC<br>AGCTTCATAACTTC      | A | T | Asn<br>(825) | Tyr<br>(825) | NON-<br>CONSER-<br>VATIVE | UNCLASSIFI-<br>ED | Human Gene SPTREMBL-<br>ACC:Q15840 ZINC FINGER<br>PROTEIN BASONUCLIN -<br>HOMO SAPIENS (HUMAN), 994<br>aa.                                       | 0 |          |
| 597 | cg43262121 | 938  | CCCAAACAGGAAAG<br>TCCATGGGCCJA<br>TGCCCTGACAGCA<br>GCTTCTTAACCTCC      | A | T | Asn<br>(826) | Ile<br>(826) | NON-<br>CONSER-<br>VATIVE | UNCLASSIFI-<br>ED | Human Gene SPTREMBL-<br>ACC:Q15840 ZINC FINGER<br>PROTEIN BASONUCLIN -<br>HOMO SAPIENS (HUMAN), 994<br>aa.                                       | 0 |          |

|     |            |      |   |   |   |     |              |                           |                   |  |                       |
|-----|------------|------|---|---|---|-----|--------------|---------------------------|-------------------|--|-----------------------|
| 598 | cg44024279 | 501  | CTGGAAAGAACCTT<br>GCCATGAGAAAGI<br>A/G]AATTTGGAG<br>AAGTACGGACATT<br>CA     | A | G | Glü | Gly<br>(827) | NON-<br>CONSER-<br>VATIVE | UNCLASSIFI-<br>ED | Human Gene SWISSPROT-<br>ACC:P0271 ALPHA-<br>FETOPROTEIN PRECURSOR<br>(ALPHA-FETOGLLOBULIN)<br>(ALPHA-1- FETOPROTEIN) -<br>Homo sapiens (Human), 609 aa.           | 0                     |
| 599 | cg44928804 | 1235 | AATGATTAAACAAAC<br>AACCTGAGACAC[G<br>/A]CGGATGAAATG<br>TTCCTGGAAACCACG<br>T | G | A | Ala | Thr<br>(828) | NON-<br>CONSER-<br>VATIVE | UNCLASSIFI-<br>ED | Human Gene SWISSPROT-<br>ACC:P21589 5'-NUCLEOTIDASE<br>PRECURSOR (EC:3.1.3.5)<br>(ECTO-NUCLEOTIDASE) (5'-<br>NT) (CD73 ANTIGEN) - Homo<br>sapiens (Human), 574 aa. | 9.1e-313<br>6 (Eq14)  |
| 600 | cg43317253 | 367  | GCCCCCAGGCCATG<br>GCTAGCTCGTGT[G/<br>T]CCGTGCAGGTGA<br>AGCTGGAGCTGGG        | G | T | Ala | Ser<br>(829) | NON-<br>CONSER-<br>VATIVE | UNCLASSIFI-<br>ED | Human Gene SWISSNEW-<br>ACC:P42568 AF-9 PROTEIN -<br>Homo sapiens (Human), 568 aa.   | 2.00E-301<br>9        |
| 601 | cg41637661 | 223  | CAGCTTTCATCCA<br>TITIATATAT[G/A]<br>GACATACTGCTAG<br>TGAAAAGACCTA           | G | A | Gly | Arg<br>(830) | NON-<br>CONSER-<br>VATIVE | UNCLASSIFI-<br>ED | Human Gene SWISSNEW-<br>ACC:Q43913 ORIGIN<br>RECOGNITION COMPLEX<br>SUBUNIT 5 - Homo sapiens<br>(Human), 435 aa.   | 6.10E-236             |
| 602 | cg42913861 | 3034 | CAGGTGTCTGCG<br>AGCCACCCGGGI<br>A/C/TCCGGGTGGC<br>GGGGTGGCGGGCG<br>GC       | A | C | Ser | Ala<br>(831) | NON-<br>CONSER-<br>VATIVE | UNCLASSIFI-<br>ED | Human Gene SWISSPROT-<br>ACC:P09529 INHIBIN BETA B<br>CHAIN PRECURSOR (ACTIVIN<br>BETA-B CHAIN) - Homo sapiens<br>(Human), 407 aa                                  | 3.00E-227<br>2 (2cen) |

|     |            |     |   |   |   |     |              |                           |                   |  |           |    |
|-----|------------|-----|---|---|---|-----|--------------|---------------------------|-------------------|--|-----------|----|
| 603 | cg43249389 | 526 | AGAGGAGAGAGGCC<br>GCCCTCGAGCGGA<br>/G/GCAAGGCGATT<br>GAGAAAAACCTCA<br>A | A | G | Ser | Gly<br>(832) | NON-<br>CONSER-<br>VATIVE | UNCLASSIFI-<br>ED | Human Gene SWISSPROT-<br>ACC:P09471 GUANINE<br>NUCLEOTIDE-BINDING<br>PROTEIN G(O), ALPHA<br>SUBUNIT 1 - Homo sapiens<br>(Human), 353 aa.   | 1.40E-188 | 15 |
| 604 | cg43919239 | 335 | GGCAGAGTTGCAG<br>CATCAGGGCCAGA<br>/C/C/TGAGCAGGAG<br>ACCCCCA/GTCCC<br>T | A | C | Ser | Arg<br>(833) | NON-<br>CONSER-<br>VATIVE | UNCLASSIFI-<br>ED | Human Gene Homologous to<br>SWISSPROT-ACC:P14207<br>FOLATE RECEPTOR BETA<br>PRECURSOR (FR-BETA)<br>(FOLATE RECEPTOR 2)<br>(FOLATE RECEPTOR,<br>FETAL/PLACENTAL)<br>(PLACENTAL FOLATE-<br>BINDING PROTEIN) (FBP) -<br>Homo sapiens (Human), 255 aa. | 4.20E-150 |    |
| 605 | cg41642952 | 787 | TAGGAATGACAGC<br>AGTAGCAGTAATA<br>/G/GGAAGGCCAA<br>AA/TCCCCCTGGAG<br>A  | A | G | Arg | Gly<br>(834) | NON-<br>CONSER-<br>VATIVE | UNCLASSIFI-<br>ED | Human Gene Homologous to<br>SWISSPROT-ACC:P21583 STEM<br>CELL FACTOR PRECURSOR<br>(SCF) (MAST CELL GROWTH<br>FACTOR) (MGF) (C-KIT<br>LIGAND) - Homo sapiens<br>(Human), 273 aa.  | 3.70E-142 | 12 |

|     |            |      |   |   |   |              |              |                           |                   |  |           |    |
|-----|------------|------|---|---|---|--------------|--------------|---------------------------|-------------------|--|-----------|----|
| 606 | cg43945147 | 221  | TGTTCTGGAGCCT<br>CAATGGTACAG[G/<br>C]GTGCTCGAGAAG<br>GACAGTGTGACT       | G | C | Arg<br>(835) | Ser<br>(835) | NON-<br>CONSER-<br>VATIVE | UNCLASSIFI-<br>ED | Human Gene Homologous to<br>SWISSNEW-ACC:P08637 LOW<br>AFFINITY IMMUNOGLOBULIN<br>GAMMA FC RECEPTOR III-1<br>PRECURSOR (FC- GAMMA<br>RII) (FCRIII) (IGG FC<br>RECEPTOR III-1) (FC-GAMMA<br>RII-ALPHA) (CD16) (FCR-10) -<br>Homo sapiens (Human), 254 aa. | 1.60E-134 | 1  |
| 607 | cg43926002 | 391  | GGGCACAGAACAC<br>CAGCACGGGGAGI<br>CS]AGCAAACACCA<br>GCACTGCCAACAG<br>AT | C | S | Ser<br>???   | ???(836)     | NON-<br>CONSER-<br>VATIVE | UNCLASSIFI-<br>ED | Human Gene Homologous to<br>SWISSPROT-ACC:P30539 MAX<br>INTERACTING PROTEIN 1<br>(MXI1 PROTEIN) - Homo sapiens<br>(Human), 228 aa.   | 1.60E-116 | 10 |
| 608 | cg43972311 | 1609 | ATTGCCATTGGGGT<br>AACTCTGGGCTCT/G<br>JCATCATCTTCAGT<br>GCCCAAATTGTG     | T | G | Glu<br>(837) | Ala<br>(837) | NON-<br>CONSER-<br>VATIVE | UNCLASSIFI-<br>ED | Human Gene Similar to<br>TREMBLNEW-ACC:AAD38008<br>GLYOXALASE-I (EC 4.4.1.5)-<br>HOMO SAPIENS (HUMAN), 184<br>aa.  | 2.20E-98  | 6  |
| 609 | cg42556108 | 521  | GTGAAGCGGTGTA<br>TGGGGACAGTG[A/C<br>/A]CCTCAACCCAGG<br>CCAGGGGGCTCCTT   | C | A | Thr<br>(838) | Ash<br>(838) | NON-<br>CONSER-<br>VATIVE | UNCLASSIFI-<br>ED | Human Gene Similar to<br>SWISSPROT-ACC:P49913<br>ANTIBACTERIAL PROTEIN<br>FALL-39 PRECURSOR (FALL-39<br>PEPTIDE ANTIBIOTIC)<br>(ANTIMICROBIAL PROTEIN<br>CAP-18) (LL-37) - Homo sapiens<br>(Human), 170 aa.  | 2.90E-87  | 3  |

|     |            |      |   |   |   |              |              |                           |                   |   |                             |
|-----|------------|------|---|---|---|--------------|--------------|---------------------------|-------------------|---|-----------------------------|
| 610 | cg36842490 | 487  | AGTGACTTCAGTA<br>AACTCTGGGTCIA/<br>CACTTTCGCCAA<br>AAAGTACCTTGAG      | A | C | Gln<br>(839) | Pro<br>(839) | NON-<br>CONSER-<br>VATIVE | UNCLASSIFI-<br>ED | Human Gene Similar to<br>SWISSPROT-ACC:P01282<br>VASOACTIVE INTESTINAL<br>PEPTIDE PRECURSOR (VIP)-<br>Homo sapiens (Human), 170 aa.       | 2.30E-85                    |
| 611 | cg43942549 | 1052 | CGGTATAACGTCA<br>AAAATCCTGTTTG/<br>TTCAGCCAAGGT<br>CAGAAATTGCCTC      | G | T | Val<br>(840) | Phe<br>(840) | NON-<br>CONSER-<br>VATIVE | UNCLASSIFI-<br>ED | Human Gene Similar to<br>SPTREMBL-ACC:Q94218<br>CODED FOR BY C. ELEGANS<br>CDNA CM10H5 -<br>CAENORHABDTIS ELEGANS,<br>589 aa.             | 2.80E-73                    |
| 612 | cg42381630 | 283  | AAGGGCTATGTA<br>CAGCCCTCTGAA[A<br>/GTGATIGGGCT<br>ATGGGGCCGAGC<br>A   | A | G | Met<br>(841) | Val<br>(841) | NON-<br>CONSER-<br>VATIVE | UNCLASSIFI-<br>ED | Human Gene Similar to<br>SPTREMBL-ACC:Q76087<br>GAGE-8 -HOMO SAPIENS<br>(HUMAN), 117 aa.  | 5.90E-64                    |
| 613 | cg42381630 | 505  | TGAAGATGGCT<br>GATGGGCAGGAGI<br>A/GTGGACCCGCC<br>AAATCCAGGGAG<br>GT   | A | G | Met<br>(842) | Val<br>(842) | NON-<br>CONSER-<br>VATIVE | UNCLASSIFI-<br>ED | Human Gene Similar to<br>SPTREMBL-ACC:Q76087<br>GAGE-8 -HOMO SAPIENS<br>(HUMAN), 117 aa.  | 5.90E-64                    |
| 614 | cg3004395  | 260  | ATTACTGAAGGT<br>GGAGAACAGAAGI<br>G/CIGTCATGAAAA<br>ATATATGCTTCAT<br>T | G | C | Gly<br>(843) | Arg<br>(843) | NON-<br>CONSER-<br>VATIVE | UNCLASSIFI-<br>ED | Human Gene Similar to<br>SPTREMBL-ACC:G238693 T<br>CELL RECEPTOR VARIABLE<br>ALPHA CHAIN - HOMO<br>SAPIENS (HUMAN), 143 aa<br>(fragment). | 1.00E-59<br>(14q11.2)<br>14 |

|     |            |     |  |   |   |     |              |                           |                   |   |          |
|-----|------------|-----|--|---|---|-----|--------------|---------------------------|-------------------|---|----------|
| 615 | cg43960645 | 733 | CACTTCCTTCT<br>CTTGGATGCCAT<br>[C]ACCCCTCTGTG<br>GGGGCGAGATGG          | A | T | Val | Glu<br>(844) | NON-<br>CONSER-<br>VATIVE | UNCLASSIFI-<br>ED | Human Gene Similar to<br>SWISSNEW-ACC:O76070<br>GAMMA-SYNUCLEIN<br>(PERSYN) (BREAST CANCER-<br>SPECIFIC GENE 1 PROTEIN)-<br>Homo sapiens (Human), 127 aa. | 1.20E-58 |
| 616 | cg2526759  | 289 | GAAGACAAGGTGG<br>TACAAAGCCCTCT<br>[C]ATCTCTGGTTGT<br>CCACGAGGGAGAC     | T | A | Leu | Gln<br>(845) | NON-<br>CONSER-<br>VATIVE | UNCLASSIFI-<br>ED | Human Gene Similar to<br>REMTREMBL-ACC:G33509 T<br>CELL RECEPTOR - HOMO<br>SAPIENS (HUMAN), 118 aa<br>(fragment).   | 1.60E-54 |
| 617 | cg2526759  | 342 | TGTAACTCTCAATT<br>GCAGTTATGAA[G]<br>ATGACTAATTC<br>GAAGCCTACTATG       | G | A | Val | Met<br>(846) | NON-<br>CONSER-<br>VATIVE | UNCLASSIFI-<br>ED | Human Gene Similar to<br>REMTREMBL-ACC:G33509 T<br>CELL RECEPTOR - HOMO<br>SAPIENS (HUMAN), 118 aa<br>(fragment).   | 1.60E-54 |
| 618 | cg2526759  | 364 | GAAGTGACTAACT<br>TTCGAAGGCCACT[T]<br>[C]ATGGTACAAGCA<br>GGAAAAGAAAGCT  | T | A | Leu | Gln<br>(847) | NON-<br>CONSER-<br>VATIVE | UNCLASSIFI-<br>ED | Human Gene Similar to<br>REMTREMBL-ACC:G33509 T<br>CELL RECEPTOR - HOMO<br>SAPIENS (HUMAN), 118 aa<br>(fragment).   | 1.60E-54 |
| 619 | cg2526759  | 475 | AGCATATTAGATA<br>AGAAAAGAACTTT[C]<br>[C]CAGGCATCCCTGA<br>ACATCACAGCCAC | T | C | Phe | Ser<br>(848) | NON-<br>CONSER-<br>VATIVE | UNCLASSIFI-<br>ED | Human Gene Similar to<br>REMTREMBL-ACC:G33509 T<br>CELL RECEPTOR - HOMO<br>SAPIENS (HUMAN), 118 aa<br>(fragment).   | 1.60E-54 |

|     |             |      |   |     |     |     |              |                |          |  |           |                      |
|-----|-------------|------|---|-----|-----|-----|--------------|----------------|----------|--|-----------|----------------------|
| 620 | cgg0310734  | 1067 | TACAGAATAATGTC<br>GTCGGTCCCC[ga<br>p]C[ACTTGAGCTG<br>GACCCTGGGAGCG<br>G | gap | C   | Thr | His<br>(849) | FRAMES<br>HIFT | cadherin | Human Gene SWISSPROT-<br>ID:P08514 PLATELET<br>MEMBRANE GLYCOPROTEIN<br>IB PRECURSOR (GP1IB)<br>(INTEGRIN ALPHA- IB) (CD41)<br>- HOMO SAPIENS (HUMAN),<br>1039 aa.   | 0         | 17<br>(17q21.3<br>2) |
| 621 | cgg0310734  | 3285 | GTCGGCTCTCAA<br>GGGAAACCGGC[ga<br>p]AJCACCCCTGGAA<br>GAAGATGATGAAG<br>A | gap | A   | Pro | His<br>(850) | FRAMES<br>HIFT | cadherin | Human Gene SWISSPROT-<br>ID:P08514 PLATELET<br>MEMBRANE GLYCOPROTEIN<br>IB PRECURSOR (GP1IB)<br>(INTEGRIN ALPHA- IB) (CD41)<br>- HOMO SAPIENS (HUMAN),<br>1039 aa.   | 0         | 17<br>(17q21.3<br>2) |
| 622 | cgg13956660 | 2521 | GTCCATCACTCAC<br>TTCAAGTTATTCT[ga<br>p]CCTAGGAGGTG<br>TATAGTCTCTGA      | T   | gap | Arg | Glu<br>(851) | FRAMES<br>HIFT | cadherin | Human Gene SWISSNEW-<br>ID:Q08722 LEUKOCYTE<br>SURFACE ANTIGEN CD47<br>PRECURSOR (ANTIGENIC<br>SURFACE DETERMINANT<br>PROTEIN OA3) (INTEGRIN<br>ASSOCIATED PROTEIN) (IAP)<br>(MER6) - HOMO SAPIENS<br>(HUMAN), 323<br>aa. Ipcis:SWISSPROT-ID:Q08722<br>LEUKOCYTE SURFACE<br>ANTIGEN CD47 PRECURSOR<br>(ANTIGENIC SURFACE<br>DETERMINANT PROTEIN OA3)<br>(INTEGRIN ASSOCIATED<br>PROTEIN) (IAP) (MER6)-<br>HOMO SAPIENS (HUMAN), 323<br>aa. | 1.80E-157 |                      |

|     |            |      |   |     |   |              |                |              |  |           |               |
|-----|------------|------|---|-----|---|--------------|----------------|--------------|--|-----------|---------------|
| 623 | cg43970982 | 2429 | CTCCAGGGATAGT<br>TGGACAGAAGGGG<br>ap/GAGACCTGGC<br>TACCCAGGACCAG<br>CT    | gap | G | Gly<br>(852) | FRAMES<br>HIFT | collagen     | Human Gene SWISSPROT-<br>ID:P12111 COLI AGEN ALPHA<br>3(VI) CHAIN PRECURSOR -<br>HOMO SAPIENS (HUMAN),<br>3176 aa.   | 0         | 2             |
| 624 | cg42175288 | 1837 | GCTATGGAGGCAA<br>AATGGGAGGAAGG<br>ap/G]AAAAGCACTAC<br>AGAAATGATCAGC<br>GC | gap | G | Arg<br>(853) | FRAMES<br>HIFT | dna_ma_bind  | Human Gene SPTREMBL-<br>ID:Q92804 PUTATIVE RNA<br>BINDING PROTEIN RBP56 -<br>HOMO SAPIENS (HUMAN), 592<br>aa.  | 0         | 17            |
| 625 | cg42175288 | 263  | CGGTIACTCCAGTT<br>ATGGACAAAGT[ga<br>p]CITATTCAACAGTC<br>CIATGGTGGTATG     | gap | C | Tyr<br>(854) | FRAMES<br>HIFT | dna_ma_bind  | Human Gene SPTREMBL-<br>ID:Q92804 PUTATIVE RNA<br>BINDING PROTEIN RBP56 -<br>HOMO SAPIENS (HUMAN), 592<br>aa.  | 0         | 17            |
| 626 | cg41554010 | 584  | GGCCGAGCAGCTG<br>CGGCCAGCTG<br>ap/G]ACCCCTACG<br>CACAGCGCATGGA<br>GA      | gap | G | Thr<br>(855) | FRAMES<br>HIFT | epiph        | Human Gene SWISSNEW-<br>ID:P06727 APOLIPOPROTEIN A-<br>IV PRECURSOR (APO-AIV) -<br>HOMO SAPIENS (HUMAN), 396<br>aa. pcis:SWISSPROT-ID:P06727<br>APOLIPOPROTEIN A-IV<br>PRECURSOR (APO-AIV) -<br>HOMO SAPIENS (HUMAN), 396<br>aa. | 1.80E-203 | 11<br>(11q23) |
| 627 | cg43065549 | 1553 | CAGACTTCCACAG<br>AGTGCTGGATGAI<br>ap/A]CGCGGCCTGC<br>CTGGCCCCAGGGTT<br>A  | gap | A | Thr<br>(856) | FRAMES<br>HIFT | glycoprotein | Human Gene SWISSPROT-<br>ID:P16452 ERYTHROCYTE<br>MEMBRANE PROTEIN BAND<br>4.2 (P4.2) (PALLIDIN) - HOMO<br>SAPIENS (HUMAN), 690 aa.  | 0         | 15<br>(15q15) |

|     |            |      |  |     |   |              |                |              |   |           |                     |
|-----|------------|------|--|-----|---|--------------|----------------|--------------|---|-----------|---------------------|
| 628 | cg41568631 | 999  | TGACCAACGGGTG<br>CTGGATGCCCTGC <sup>g</sup><br>TCCTTATACATCCT<br>GGACCGGGGGGG<br>A                             | gap | C | Leu<br>(857) | FRAMES<br>HIFT | glycoprotein | Human Gene Similar to<br>SWISSPROT-ID:P16452<br>ERYTHROCYTE MEMBRANE<br>PROTEIN BAND 4.2 (P <sup>4.2</sup> )<br>(PALLIDIN) - HOMO SAPIENS<br>(HUMAN), 690 aa. | 9.90E-70  | 14<br>(14q11.2<br>) |
| 629 | cg41637704 | 1220 | GCGCCGGAGACA<br>AGGGCAGCGGGAC <sup>b</sup><br>ap/GJGCCTGCGGA<br>CTTGAGGGACAGT<br>GA                            | gap | G | Pro<br>(858) | FRAMES<br>HIFT | homeobox     | Human Gene SWISSPROT-<br>ID:P50219 HOMEOBOX<br>PROTEIN HB9 - HOMO<br>SAPIENS (HUMAN), 401 aa.   | 1.20E-224 | 7                   |
| 630 | cg43933380 | 364  | ATAAGTTACAATG<br>CTTITTTGTTTA <sup>g</sup><br>ap AAA <sup>a</sup> AAA <sup>a</sup> AAA<br>AACTCTGTACTTTA<br>GA | gap | A | Leu<br>(859) | FRAMES<br>HIFT | interferon   | Human Gene SWISSPROT-<br>ID:P15260 INTERFERON-<br>GAMMA RECEPTOR ALPHA<br>CHAIN PRECURSOR (CDW119)<br>- HOMO SAPIENS (HUMAN),<br>489 aa.                      | 1.40E-261 | 6                   |
| 631 | cg43072541 | 379  | CTGTGGGCGGGT<br>TCTGTATCTGAT <sup>g</sup><br>pCJATCATTCGATT<br>ACGAAATAAACG<br>T                               | gap | C | Ile<br>(860) | FRAMES<br>HIFT | kinase       | Human Gene SPTRMBL-<br>ID:Q15802 SERINE/THREONINE<br>PROTEIN KINASE KRS-2-<br>HOMO SAPIENS (HUMAN), 487<br>aa.  | 9.60E-262 | 20                  |

|     |             |      |  |     |   |     |              |                |                  |  |           |          |
|-----|-------------|------|--|-----|---|-----|--------------|----------------|------------------|--|-----------|----------|
| 632 | cgt4032168  | 1536 | GTCAGCCGCTACC<br>TCGACTGGATCC<br>p/TJATGGCACATC<br>AGAGACAAGGAAG<br>C    | gap | T | His | Leu<br>(861) | FRAMES<br>HIFT | protease         | Human Gene Similar to<br>SWISSPROT-ID:P25155<br>COAGULATION FACTOR X<br>PRECURSOR (EC 3.4.21.6)<br>(STUART FACTOR) (VIRUS<br>ACTIVATING PROTEASE)<br>(VAP) - GALLUS GALLUS<br>(CHICKEN), 475 aa. | 2.40E-82  | 2 (2q13) |
| 633 | cgt43931248 | 1317 | CCGGGCCAGAGCTG<br>CGTCTGCTGAGG<br>ap/GJCTCAAGTTAA<br>AAGTGGAGCAGCA<br>CG | gap | G | Leu | Ala<br>(862) | FRAMES<br>HIFT | tgf <sup>f</sup> | Human Gene SWISSPROT-<br>ID:P01137 TRANSFORMING<br>GROWTH FACTOR BETA 1<br>PRECURSOR (TGF-BETA 1)-<br>HOMO SAPIENS (HUMAN), 390<br>aa.   | 9.70E-214 | 19       |
| 634 | cgt43931248 | 1317 | CCGGGCCAGAGCTG<br>CGTCTGCTGAGG<br>ap/GJCTCAAGTTAA<br>AAGTGGAGCAGCA<br>CG | gap | G | Leu | Ala<br>(863) | FRAMES<br>HIFT | tgf <sup>f</sup> | Human Gene SWISSPROT-<br>ID:P01137 TRANSFORMING<br>GROWTH FACTOR BETA 1<br>PRECURSOR (TGF-BETA 1)-<br>HOMO SAPIENS (HUMAN), 390<br>aa.   | 9.70E-214 | 19       |
| 635 | cgt43272560 | 847  | AATCTCCGCACTG<br>CAGGCCAGGGC<br>ap/CJTGGCAGCTA<br>CAGAGAGGGTCA<br>CA     | gap | C | Ala | Ala<br>(864) | FRAMES<br>HIFT | tgf receptor     | Human Gene SWISSPROT-<br>ID:Q03167 TGF-BETA<br>RECEPTOR TYPE III<br>PRECURSOR (TGFR-3)<br>(BETAGLYCAN) - HOMO<br>SAPIENS (HUMAN), 849 aa.  | 0         | 1 (1p33) |

|     |            |      |  |     |   |              |                |             |   |           |   |
|-----|------------|------|--|-----|---|--------------|----------------|-------------|---|-----------|---|
| 636 | cg43266471 | 1067 | CCAGGATCCATT T<br>GAGGATTATGG [gap<br>T]TGCTGGACA<br>CCATCAACTCCTCA  | gap | T | Gly<br>(865) | FRAMES<br>HIFT | tmt7        | Human Gene SWISSPROT-<br>ID:P32241 VASOACTIVE<br>INTESTINAL POLYPEPTIDE<br>RECEPTOR 1 PRECURSOR<br>(VIP-R-1) (PITUITARY<br>ADENYLATE CYCLASE<br>ACTIVATING POLYPEPTIDE<br>TYPE II RECEPTOR) (PACAP<br>TYPE II RECEPTOR) (PACAP-R-<br>2) - HOMO SAPIENS (HUMAN),<br>457 aa.                      | 5.20E-254 | 3 |
| 637 | cg43995237 | 625  | CAAATCCCCCGTT<br>TCTTCATCTTG [gap/<br>GIACATGCTAAAT<br>GAAATTACGCACT | gap | G | Gln<br>(866) | FRAMES<br>HIFT | transferase | Human Gene SWISSPROT-<br>ID:P53611 GERANYLGERANYL<br>TRANSFERASE TYPE II BETA<br>SUBUNIT (EC 2.5.1.-) (RAB<br>GERANYLGERANYL TRANSFE-<br>RASE BETA SUBUNIT) (RAB<br>GERANYL-<br>GERANYL TRANSFERASE<br>BETA SUBUNIT) (RAB GG<br>TRANSFERASE) (RAB<br>GGTASE) - HOMO SAPIENS<br>(HUMAN), 331 aa. | 1.40E-182 | 1 |

|     |            |     |  |     |              |                |                 |  |  |           |   |
|-----|------------|-----|--|-----|--------------|----------------|-----------------|--|--|-----------|---|
| 638 | cg43995237 | 638 | TTCCTCATCTTGAC<br>ATGCTAAAATG[gap]<br>/GIAAAATTACGCAG<br>TTTCTCTCTATCAA        | gap | G            | Leu<br>(867)   | FRAMES<br>HIFT  | transferase  | Human Gene SWISSPROT-<br>ID:P53611 GERANYLGERANYL-<br>TRANSFERASE TYPE II BETA<br>SUBUNIT (EC 2.5.1.-) (RAB<br>GERANYLGERANYLTRANSFE-<br>RASE BETA SUBUNIT) (RAB<br>GERANYL-<br>GERANYLTRANSFERASE<br>BETA SUBUNIT) (RAB GG<br>TRANSFERASE) (RAB<br>GGTASE) - HOMO SAPIENS<br>(HUMAN), 331 aa. | 1.40E-182 | 1 |
| 639 | cg43254094 | 267 | CCGCCTCTGCTGCT<br>GCTGCTGCTGCTGCTGCTGCT<br>/GIGCGTCCCCGCC<br>AGCCGAGCTTCC<br>C | gap | G            | Arg<br>(868)   | FRAMES<br>HIFT  | UNCLASSIFI<br>D  | Human Gene SWISSPROT-<br>ACC:P78539 SUSHI REPEAT-<br>CONTAINING PROTEIN SRPX<br>PRECURSOR - Homo sapiens<br>(Human), 464 aa.   | 6.40E-257 | X |
| 640 | cg44034555 | 665 | ATCCAGGGCTGAGC<br>TGGATCATCTGA[G<br>/gap]GGCCCTCCAGC<br>CACCCGTTTCCCT<br>T     | gap | Pro          | Leu<br>(869)   | FRAMES<br>HIFT  | UNCLASSIFI<br>D  | Human Gene SWISSNEW-<br>ACC:Q13228 SELENTUM-<br>BINDING PROTEIN 1 - Homo<br>sapiens (Human), 472 aa.   | 3.80E-252 | 1 |
| 641 | cg44034555 | 667 | CCAGGGCTGAGCTG<br>GATCATCTGAGG[G<br>/gap]CCTCCAGCCA<br>CCCGTTTCCCTG<br>A       | gap | Gly<br>(870) | FRAMES<br>HIFT | UNCLASSIFI<br>D | Human Gene SWISSNEW-<br>ACC:Q13228 SELENTUM-<br>BINDING PROTEIN 1 - Homo<br>sapiens (Human), 472 aa. | 3.80E-252  | 1         |   |

|     |            |     |   |     |              |              |              |                |                   |  |           |               |
|-----|------------|-----|---|-----|--------------|--------------|--------------|----------------|-------------------|--|-----------|---------------|
| 642 | cg39711096 | 882 | AGCGGAGTCCTCCG<br>GGAGGCCACAG[G<br>ap]GTTTACAGCCTC<br>CAGCTGCAGCACT<br>GA | gap | G            | Val<br>(871) | Gly<br>(871) | FRAMES<br>HIFT | UNCLASSIFI[E<br>D | Human Gene SWISSPROT-<br>ACC:P18428<br>LIPOPOLYSACCHARIDE-<br>BINDING PROTEIN<br>PRECURSOR (LBP) - Homo<br>sapiens (Human), 481 aa.  | 1.00E-251 |               |
| 643 | cg44128902 | 379 | CGTICCAAGGGAG<br>CATATCTGCTGAG[A<br>p]CTGATGACCTGCA<br>AAGAGTCATCCAG<br>A | gap | C            | Asp<br>(872) | Asp<br>(872) | FRAMES<br>HIFT | UNCLASSIFI[E<br>D | Human Gene SWISSPROT-<br>ACC:P18615 RD PROTEIN -<br>Homo sapiens (Human), 380 aa.  | 1.00E-201 | 1<br>(1p36.2) |
| 644 | cg43946951 | 306 | GGAACTGGCAC<br>GTCGTCGGGGAG[C<br>/gap]CCCCAAGATCA<br>CCGGCGCCCTGT<br>GT   | gap | Gly<br>(873) | Gly<br>(873) | Gly<br>(873) | FRAMES<br>HIFT | UNCLASSIFI[E<br>D | Human Gene SWISSPROT-<br>ACC:P09467 FRUCTOSE-1,6-<br>BISPHOSPHATASE (EC 3.1.3.11)<br>(D-FRUCTOSE-1,6-<br>BISPHOSPHATE 1-<br>PHOSPHOHYDROLASE)<br>(FBPASE) - Homo sapiens<br>(Human), 337 aa. | 3.50E-178 | 9<br>(9q22.2) |
| 645 | cg43948890 | 195 | ATTCCCGGGGAG<br>GGGGCCCTGTAAG[G<br>/gap]GGAAACAGA<br>CAATCCCATGAGA<br>CT  | gap | Pro<br>(874) | Leu<br>(874) | Leu<br>(874) | FRAMES<br>HIFT | UNCLASSIFI[E<br>D | Human Gene Homologous to<br>SPTREMBL-ACC:Q15182<br>SNRNP POLYPEPTIDE B -<br>HOMO SAPIENS (HUMAN), 285<br>aa.   | 3.20E-147 | 20            |
| 646 | cg43948890 | 197 | TCCCGGGGAGGG<br>GGCCCTGTAAGG[G<br>/gap]AAACAGACA<br>AICCAGAGACT<br>CC     | gap | Phe<br>(875) | Phe<br>(875) | Phe<br>(875) | FRAMES<br>HIFT | UNCLASSIFI[E<br>D | Human Gene Homologous to<br>SPTREMBL-ACC:Q15182<br>SNRNP POLYPEPTIDE B -<br>HOMO SAPIENS (HUMAN), 285<br>aa.   | 3.20E-147 | 20            |

|     |            |     |   |     |              |     |              |                |                   |   |           |          |
|-----|------------|-----|---|-----|--------------|-----|--------------|----------------|-------------------|---|-----------|----------|
| 647 | cg43917524 | 713 | GGGCCCTGTCTGCC<br>CAGTGGAGGAGGI<br>C/gap]TCGCCGCTGGT<br>GITCTAGGGGGCA<br>TC | C   | gap          | Ala | Pro<br>(876) | FRAMES<br>HIFT | UNCLASSIFIIE<br>D | Human Gene Homologous to<br>TREMBLNEW-ACC:AAD43025<br>PTD017 - HOMO SAPIENS<br>(HUMAN), 258 aa.   | 3.20E-143 |          |
| 648 | cg43942004 | 373 | CTCTCGGCACTGGT<br>GACTGGCGAGA[ga<br>p]GICCTGGAGCGG<br>CITCGGAGAGGGC<br>TA   | gap | Glu<br>(877) | Asp |              | FRAMES<br>HIFT | UNCLASSIFIIE<br>D | Human Gene Homologous to<br>SWISSNEW-ACC:Q99075<br>HEPARIN-BINDING EGF-LIKE<br>GROWTH FACTOR<br>PRECURSOR (HB-EGF)<br>(HBEGF) (DIPHTERIA TOXIN<br>RECEPTOR) (DT-R) - Homo<br>sapiens (Human), 203 aa. | 1.00E-107 | 5 (5q23) |
| 649 | cg43932428 | 681 | TCGTGGCCAGGTC<br>CTTCTGCCTGAAG[C/<br>gap]CCCCTTGCTCTG<br>CCGACCTTGCTGG<br>A | C   | gap          | Gly | Gly<br>(878) | FRAMES<br>HIFT | UNCLASSIFIIE<br>D | Human Gene Similar to<br>SPTREMBL-ACC:Q60869 EDF-1<br>PROTEIN - HOMO SAPIENS<br>(HUMAN), 148 aa.  | 2.50E-72  |          |
| 650 | cg44010855 | 450 | GGTCCAATGCAA<br>GTGCTCCCGAAAG<br>[gap]GGACCAAGA<br>TCCGCTACAGCGA<br>CG      | G   | gap          | Gly | Asp<br>(879) | FRAMES<br>HIFT | UNCLASSIFIIE<br>D | Human Gene Similar to<br>TREMBLNEW-ACC:AAD38944<br>NIAC PROTEIN - HOMO<br>SAPIENS (HUMAN), 99 aa.   | 5.80E-50  | 5        |
| 651 | cg44010855 | 452 | TCCAATGCAAGT<br>GCTCCGGAAAG[G<br>/gap]ACCAAGATC<br>CGCTACAGCGACG<br>TG      | G   | gap          | Gly | Asp<br>(880) | FRAMES<br>HIFT | UNCLASSIFIIE<br>D | Human Gene Similar to<br>TREMBLNEW-ACC:AAD38944<br>NIAC PROTEIN - HOMO<br>SAPIENS (HUMAN), 99 aa.   | 5.80E-50  | 5        |

**CLAIMS**

WHAT IS CLAIMED IS:

1. An isolated polynucleotide selected from the group consisting of:
  - a) a nucleotide sequence comprising one or more polymorphic sequences (SEQ ID NOS:1 - 651);
    - 5 b) a fragment of said nucleotide sequence, provided that the fragment includes a polymorphic site in said polymorphic sequence;
    - c) a complementary nucleotide sequence comprising a sequence complementary to one or more of said polymorphic sequences (SEQ ID NOS:1 - 651); and
    - 10 d) a fragment of said complementary nucleotide sequence, provided that the fragment includes a polymorphic site in said polymorphic sequence.
2. The polynucleotide of claim 1, wherein said polynucleotide sequence is DNA.
  - 15
  3. The polynucleotide of claim 1, wherein said polynucleotide sequence is RNA.
  4. The polynucleotide of claim 1, wherein said polynucleotide sequence is between about 10 and about 100 nucleotides in length.
    - 20
    5. The polynucleotide of claim 1, wherein said polynucleotide sequence is between about 10 and about 90 nucleotides in length.

6. The polynucleotide of claim 1, wherein said polynucleotide sequence is between about 10 and about 75 nucleotides in length.
7. The polynucleotide of claim 1, wherein said polynucleotide is between about 10 and  
5 bases in length.
8. The polynucleotide of claim 1, wherein said polynucleotide is between about 10 and  
about 40 bases in length.
- 10 9. The polynucleotide of claim 1, wherein said polynucleotide is derived from a nucleic  
acid encoding a polypeptide related to angiopoietin, 4-hydroxybutyrate  
dehydrogenase, ATP-dependent RNA helicase, MHC Class I histocompatibility  
antigen, or phosphoglycerate kinase.
- 15 10. The polynucleotide of claim 1, wherein said polymorphic site includes a nucleotide  
other than the nucleotide listed in Table 1, column 5 for said polymorphic sequence.
11. The polynucleotide of claim 1, wherein the complement of said polymorphic site  
includes a nucleotide other than the complement of the nucleotide listed in Table 1,  
20 column 5 for the complement of said polymorphic sequence.
12. The polynucleotide of claim 1, wherein said polymorphic site includes the nucleotide  
listed in Table 1, column 6 for said polymorphic sequence.

13. The polynucleotide of claim 1, wherein the complement of said polymorphic site includes the complement of the nucleotide listed in Table 1, column 6 for said polymorphic sequence.

5 14. An isolated allele-specific oligonucleotide that hybridizes to a first polynucleotide at a polymorphic site encompassed therein, wherein the first polynucleotide is chosen from the group consisting of:

a) a nucleotide sequence comprising one or more polymorphic sequences (SEQ ID NOS:1 - 651) provided that the polymorphic sequence includes a

10 nucleotide other than the nucleotide recited in Table 1, column 5 for said polymorphic sequence;

b) a nucleotide sequence that is a fragment of said polymorphic sequence, provided that the fragment includes a polymorphic site in said polymorphic sequence;

15 c) a complementary nucleotide sequence comprising a sequence complementary to one or more polymorphic sequences (SEQ ID NOS:1 - 651), provided that the complementary nucleotide sequence includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5; and

20 d) a nucleotide sequence that is a fragment of said complementary sequence, provided that the fragment includes a polymorphic site in said polymorphic sequence.

15. The oligonucleotide of claim 14, wherein the oligonucleotide does not hybridize under stringent conditions to a second polynucleotide selected from the group consisting of:

a) a nucleotide sequence comprising one or more polymorphic sequences (SEQ ID NOS:1 - 651), wherein said polymorphic sequence includes the nucleotide listed in Table 1, column 5 for said polymorphic sequence;

5 b) a nucleotide sequence that is a fragment of any of said nucleotide sequences;

c) a complementary nucleotide sequence comprising a sequence complementary to one or more polymorphic sequences (SEQ ID NOS:1 - 651), wherein said polymorphic sequence includes the complement of the nucleotide listed in Table 1, column 5; and

10 d) a nucleotide sequence that is a fragment of said complementary sequence, provided that the fragment includes a polymorphic site in said polymorphic sequence.

16. The oligonucleotide of claim 15, wherein the oligonucleotide is between about 10 and  
15 about 51 bases in length.

17. The oligonucleotide of claim 15, wherein the oligonucleotide identifies a polypeptide related to angiopoietin, 4-hydroxybutyrate dehydrogenase, ATP-dependent RNA helicase, MHC Class I histocompatibility antigen, or phosphoglycerate kinase.  
20

18. The oligonucleotide of claim 15, wherein the oligonucleotide is between about 15 and about 30 bases in length.

19. A method of detecting a polymorphic site in a nucleic acid, the method comprising:  
25 a) contacting said nucleic acid with an oligonucleotide that hybridizes to a polymorphic sequence selected from the group consisting of SEQ ID NOS: 1 - 651, or its complement, provided that the polymorphic sequence

includes a nucleotide other than the nucleotide recited in Table 1, column 5 for said polymorphic sequence, or the complement includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5; and

5            b) determining whether said nucleic acid and said oligonucleotide hybridize; whereby hybridization of said oligonucleotide to said nucleic acid sequence indicates the presence of the polymorphic site in said nucleic acid.

20.        The method of claim 19, wherein said oligonucleotide does not hybridize to said polymorphic sequence when said polymorphic sequence includes the nucleotide recited in Table 1, column 5 for said polymorphic sequence, or when the complement of the polymorphic sequence includes the complement of the nucleotide recited in Table 1, column 5 for said polymorphic sequence.

15        21.      The method of claim 19, wherein said oligonucleotide identifies a polypeptide related to angiopoietin, 4-hydroxybutyrate dehydrogenase, ATP-dependent RNA helicase, MHC Class I histocompatibility antigen, or phosphoglycerate kinase.

20        22.      The method of claim 19, wherein said oligonucleotide is between about 15 and about 30 bases in length.

23.        A method of detecting the presence of a sequence polymorphism in a subject, the method comprising:

25            a) providing a nucleic acid from said subject;

                b) contacting said nucleic acid with an oligonucleotide that hybridizes to a polymorphic sequence selected from the group consisting of SEQ ID NOS:1 - 651, or its complement, provided that the polymorphic sequence

includes a nucleotide other than the nucleotide recited in for said polymorphic sequence, or the complement includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5; and

c) determining whether said nucleic acid and said oligonucleotide hybridize;

5 whereby hybridization of said oligonucleotide to said nucleic acid sequence indicates the presence of the polymorphism in said subject.

24. A method of determining the relatedness of a first and second nucleic acid, the method comprising:

10 a) providing a first nucleic acid and a second nucleic acid;

b) contacting said first nucleic acid and said second nucleic acid with an oligonucleotide that hybridizes to a polymorphic sequence selected from the group consisting of SEQ ID NOS:1 - 651, or its complement, provided that the polymorphic sequence includes a nucleotide other than the nucleotide recited in Table 1, column 5 for said polymorphic sequence, or the complement includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5;

15 c) determining whether said first nucleic acid and said second nucleic acid hybridize to said oligonucleotide; and

d) comparing hybridization of said first and second nucleic acids to said oligonucleotide,

20 wherein hybridization of the first and second nucleic acids to said oligonucleotide indicates the first and second nucleic acids are related.

25 25. The method of claim 24, wherein said oligonucleotide does not hybridize to said polymorphic sequence when said polymorphic sequence includes the nucleotide recited in Table 1, column 5 for said polymorphic sequence, or when the complement

of the polymorphic sequence includes the complement of the nucleotide recited in Table 1, column 5 for said polymorphic sequence.

26. The method of claim 24, wherein the oligonucleotide is between about 10 and about  
5 bases in length.
27. The method of claim 24, wherein the oligonucleotide is between about 10 and about  
40 bases in length.
- 10 28. The method of claim 24, wherein the oligonucleotide is between about 15 and about  
30 bases in length.
29. An isolated polypeptide comprising a polymorphic site at one or more amino acid  
residues, wherein the protein is encoded by a polynucleotide selected from the group  
consisting of: polymorphic sequences SEQ ID NOS:1 - 651, or their complement,  
15 provided that the polymorphic sequence includes a nucleotide other than the  
nucleotide recited in Table 1, column 5 for said polymorphic sequence, or the  
complement includes a nucleotide other than the complement of the nucleotide recited  
in Table 1, column 5.
- 20 30. The polypeptide of claim 29, wherein said polypeptide is translated in the same open  
reading frame as is a wild type protein whose amino acid sequence is identical to the  
amino acid sequence of the polymorphic protein except at the site of the  
polymorphism.
- 25 31. The polypeptide of claim 29, wherein the polypeptide encoded by said polymorphic  
sequence, or its complement, includes the nucleotide listed in Table 2, column 6 or

Table 3, column 5 for said polymorphic sequence, or the complement includes the complement of the nucleotide listed in Table 1, column 6.

32. An antibody that binds specifically to a polypeptide encoded by a polynucleotide comprising a nucleotide sequence encoded by a polynucleotide selected from the group consisting of polymorphic sequences SEQ ID NOS:1 - 651, or its complement, provided that the polymorphic sequence includes a nucleotide other than the nucleotide recited in Table 1, column 5 for said polymorphic sequence, or the complement includes a nucleotide other than the complement of the nucleotide recited in Table 1, column 5.

33. The antibody of claim 32, wherein said antibody binds specifically to a polypeptide encoded by a polymorphic sequence which includes the nucleotide listed in Table 1, column 6 for said polymorphic sequence.

34. The antibody of claim 32, wherein said antibody does not bind specifically to a polypeptide encoded by a polymorphic sequence which includes the nucleotide listed in Table 1, column 5 for said polymorphic sequence.

35. A method of detecting the presence of a polypeptide having one or more amino acid residue polymorphisms in a subject, the method comprising

- providing a protein sample from said subject;
- contacting said sample with the antibody of claim 34 under conditions that allow for the formation of antibody-antigen complexes; and
- detecting said antibody-antigen complexes,

whereby the presence of said complexes indicates the presence of said polypeptide.

36. A method of treating a subject suffering from, at risk for, or suspected of, suffering from a pathology ascribed to the presence of a sequence polymorphism in a subject, the method comprising:

5           a) providing a subject suffering from a pathology associated with aberrant expression of a first nucleic acid comprising a polymorphic sequence selected from the group consisting of SEQ ID NOS:1 - 651, or its complement; and

10          b) administering to the subject an effective therapeutic dose of a second nucleic acid comprising the polymorphic sequence, provided that the second nucleic acid comprises the nucleotide present in a wild type allele of the sequence polymorphism,

              thereby treating said subject.

15   37. The method of claim 36, wherein the second nucleic acid sequence comprises a polymorphic sequence which includes the nucleotide listed in Table 1, column 5 for said polymorphic sequence.

20   38. A method of treating a subject suffering from, at risk for, or suspected of suffering from a pathology ascribed to the presence of a sequence polymorphism in a subject, the method comprising:

25           a) providing a subject suffering from a pathology associated with aberrant expression of a polymorphic sequence selected from the group consisting of polymorphic sequences SEQ ID NOS:1 - 651, or its complement; and

              b) administering to the subject an effective therapeutic dose of a polypeptide,

wherein said polypeptide is encoded by a polynucleotide comprising a polymorphic sequence selected from the group consisting of SEQ ID NOS:1 - 651, or by a polynucleotide comprising a nucleotide sequence that is complementary to any one of polymorphic sequences SEQ ID NOS:1 - 651, provided that said polymorphic sequence includes the nucleotide listed in Table 1, column 6 for said polymorphic sequence, thereby treating said subject.

5

39. A method of treating a subject suffering from, at risk for, or suspected of suffering from, a pathology ascribed to the presence of a sequence polymorphism in a subject, the method comprising:

10

a) providing a subject suffering from, at risk for, or suspected of suffering from, a pathology associated with aberrant expression of a first nucleic acid comprising a polymorphic sequence selected from the group consisting of SEQ ID NOS:1 - 651, or its complement; and

15

b) administering to the subject an effective dose of the antibody of claim 34, thereby treating said subject.

20

40. A method of treating a subject suffering from, at risk for, or suspected of suffering from, a pathology ascribed to the presence of a sequence polymorphism in a subject, the method comprising:

25

a) providing a subject suffering from, at risk for, or suspected of suffering from, a pathology associated with aberrant expression of a nucleic acid comprising a polymorphic sequence selected from the group consisting of SEQ ID NOS:1 - 651, or its complement; and

b) administering to the subject an effective dose of an oligonucleotide comprising a polymorphic sequence selected from the group consisting of SEQ ID NOS:1 - 651, or by a polynucleotide comprising a nucleotide sequence that is complementary to any one of polymorphic sequences SEQ

ID NOS:1 - 651, provided that said polymorphic sequence includes the nucleotide listed in Table 1, column 6 for said polymorphic sequence,

thereby treating said subject.

5 41. An oligonucleotide array, comprising one or more oligonucleotides hybridizing to a first polynucleotide at a polymorphic site encompassed therein, wherein the first polynucleotide is chosen from the group consisting of:

a) a nucleotide sequence comprising one or more polymorphic sequences SEQ

ID NOS:1 - 651;

10 b) a nucleotide sequence that is a fragment of any of said nucleotide sequence, provided that the fragment includes a polymorphic site in said polymorphic sequence;

c) a complementary nucleotide sequence comprising a sequence complementary to one or more polymorphic sequences SEQ ID NOS:1 -

15 651; and

d) a nucleotide sequence that is a fragment of said complementary sequence, provided that the fragment includes a polymorphic site in said polymorphic sequence.

20 42. The array of claim 41, wherein said array comprises 10 oligonucleotides.

43. The array of claim 41, wherein said array comprises at least 100 oligonucleotides.

44. The array of claim 41, wherein said array comprises at least 1000 oligonucleotides.

1/1

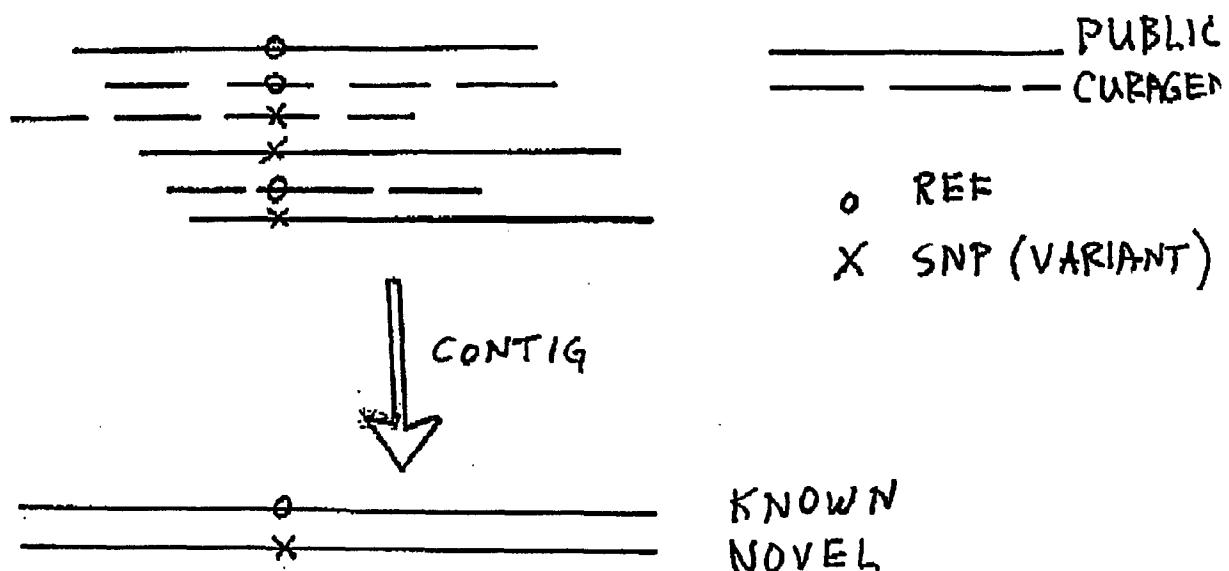


FIG. 1

## SEQUENCE LISTING

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Shimkets, Richard A.  
Leach, Martin D.

<120> Nucleic Acids Containing Single Nucleotide Polymorphisms and Methods of Use Thereof

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<141> 2000-12-27

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&lt;212&gt; DNA

&lt;213&gt; Homo sapiens

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&lt;221&gt; allele

&lt;222&gt; (26)...(0)

&lt;223&gt; single nucleotide polymorphism

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&lt;222&gt; (0)...(0)

&lt;223&gt; Accession number cg42659872

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51

&lt;210&gt; 68

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&lt;223&gt; single nucleotide polymorphism

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&lt;222&gt; (0)...(0)

&lt;223&gt; Accession number cg42506800

&lt;400&gt; 68

GCTTGCCAAT TTCTCGTCTG TATGCCAAGT ACTTTCAAGG AGATCTGAAT C

51

&lt;210&gt; 69

&lt;211&gt; 51

&lt;212&gt; DNA

&lt;213&gt; Homo sapiens

&lt;220&gt;

&lt;221&gt; allele

&lt;222&gt; (26)...(0)

&lt;223&gt; single nucleotide polymorphism

&lt;221&gt; misc\_feature

&lt;222&gt; (0)...(0)

&lt;223&gt; Accession number cg43966621

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&lt;211&gt; 51

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<210> 653  
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<213> Homo sapiens

<220>  
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<223> cSNP translation

<400> 653

Arg Leu His Arg Leu Arg Gly Glu Gln Met Ala Ser Tyr Phe  
1 5 10

<210> 654  
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<220>  
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<222> (7)...(0)  
<223> cSNP translation

<400> 654

Gly Ala Pro Leu Tyr Met Asp Ser Arg Ala Asp Arg Lys Leu  
1 5 10

<210> 655  
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<223> cSNP translation

<400> 655

Gln Ala Gly Thr Thr Leu Asp Leu Asp Leu Gly Gly Lys His  
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<223> cSNP translation

<400> 656

Lys Cys Glu Cys Ser Arg Ala Tyr Gln Met Asp Leu Ala Thr  
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<210> 657

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<223> cSNP translation

<400> 657

Asp Val Arg Gly Asn Leu Lys Gly Asn Thr Glu Gly Leu Gln  
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<210> 658

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<223> cSNP translation

<400> 658

Val Pro Leu Glu Thr Pro Arg Val His Ser Arg Ala Pro Ser  
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<223> cSNP translation

<400> 659

Pro Phe Gly Val Ile Val Arg Arg Gln Leu Asp Gly Arg Val  
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<210> 660

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<400> 660

Trp Gly Lys Glu Pro Lys Leu Glu Ser Ala Trp Met Asn Gly  
1 5 10

<210> 661

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<223> cSNP translation

<400> 661

Ser Arg Gly Tyr Arg Asn Arg Arg Ser Ser Arg Glu Thr Arg  
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<210> 662

<211> 14

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<213> Homo sapiens

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<222> (7)...(0)

<223> cSNP translation

<400> 662

Gly Ala Met Leu Leu Asn Ile Ser Gly His Val Lys Glu Ser  
1 5 10

<210> 663

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<212> PRT

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<222> (7)...(0)

<223> cSNP translation

<400> 663

Gln Gly Gly Lys Leu Ser Val Val Leu Arg Ala Glu Asp Ile  
1 5 10

<210> 664

<211> 14

<212> PRT

<213> Homo sapiens

<220>  
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<223> cSNP translation

<400> 664  
Pro Gly Asp Ile Ser Arg Leu Leu Glu Phe Thr Lys Ala His  
1 5 10

<210> 665  
<211> 14  
<212> PRT  
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<220>  
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Val Val Ile Pro Ser Asp Phe Phe Gln Ile Val Gly Gly Ser  
1 5 10

<210> 666  
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<400> 666  
Lys Arg Lys Leu Phe Ile Arg Ser Met Gly Glu Gly Thr Ile  
1 5 10

<210> 667  
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<213> Homo sapiens

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<400> 667  
Glu Gln Ala Arg Gln Gly Leu Lys Gly Leu Glu Glu Thr Val  
1 5 10

<210> 668  
<211> 14  
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<400> 668  
Cys  
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<400> 669  
Ser Phe Leu Ile Ser Pro Leu Thr Pro Ala His Ala Gly Thr  
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<210> 670  
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<400> 670  
Trp Cys Ser Lys Lys Asp Ala Ala Val Met Asn Gln Glu  
1 5 10

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<400> 671  
Val Ala Leu Pro Gln Pro Leu Gly Val Pro Asn Glu Ser Gln  
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<210> 672  
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<400> 672  
Gly Ile Gln Asn Lys Glu Val Glu Val Arg Ile Phe His Cys  
1 5 10

<210> 673  
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<222> (7)...(0)  
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<400> 673  
Val Asp Asn Ile Arg Ser Val Phe Gly Asn Ala Val Ser Arg  
1 5 10

<210> 674  
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<220>  
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<400> 674  
Leu Thr Ile Gln Leu Ile Glu Asn His Phe Val Asp Glu Tyr  
1 5 10

<210> 675  
<211> 14  
<212> PRT  
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<220>  
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<400> 675  
Tyr Gly Ile Pro Phe Ile Gln Thr Ser Ala Lys Thr Arg Gln  
1 5 10

<210> 676  
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<212> PRT  
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<220>  
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<400> 676  
Ser Ala Ser Lys Gln Ala Val Arg Pro Val Leu Ala Thr Thr  
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<210> 677  
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<212> PRT  
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<220>  
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<223> cSNP translation

<400> 677  
Tyr Cys Met Val Phe Leu Ala Leu Tyr Val Gln Ala Arg Leu  
1 5 10

&lt;210&gt; 678

&lt;211&gt; 14

&lt;212&gt; PRT

&lt;213&gt; Homo sapiens

&lt;220&gt;

&lt;221&gt; VARIANT

&lt;222&gt; (7)...(0)

&lt;223&gt; cSNP translation

&lt;400&gt; 678

Val Gly Thr Tyr Arg Cys Val Pro Gly Lys Lys Gly Gly Tyr  
1 5 10

&lt;210&gt; 679

&lt;211&gt; 14

&lt;212&gt; PRT

&lt;213&gt; Homo sapiens

&lt;220&gt;

&lt;221&gt; VARIANT

&lt;222&gt; (7)...(0)

&lt;223&gt; cSNP translation

&lt;400&gt; 679

Gly Arg Ala Thr Ser Gly Ser Glu His Gln Phe Cys Gly Gly  
1 5 10

&lt;210&gt; 680

&lt;211&gt; 14

&lt;212&gt; PRT

&lt;213&gt; Homo sapiens

&lt;220&gt;

&lt;221&gt; VARIANT

&lt;222&gt; (7)...(0)

&lt;223&gt; cSNP translation

&lt;400&gt; 680

Gly Glu Trp Ile Thr Val Asp Gln Thr Thr Ala Asn Arg  
1 5 10

&lt;210&gt; 681

&lt;211&gt; 14

&lt;212&gt; PRT

&lt;213&gt; Homo sapiens

&lt;220&gt;

&lt;221&gt; VARIANT

&lt;222&gt; (7)...(0)

&lt;223&gt; cSNP translation

&lt;400&gt; 681

Pro Glu Leu Val Leu Glu Leu Pro Ile Arg His Pro Lys Phe  
1 5 10

&lt;210&gt; 682

&lt;211&gt; 14

&lt;212&gt; PRT

&lt;213&gt; Homo sapiens

&lt;220&gt;

&lt;221&gt; VARIANT

&lt;222&gt; (7)...(0)

<223> cSNP translation

<400> 682

Glu Trp Phe Lys Asp Leu Ala Leu Lys Trp Tyr Gly Leu Pro  
1 5 10

<210> 683

<211> 14

<212> PRT

<213> Homo sapiens

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<223> cSNP translation

<400> 683

Tyr Ile Thr Gly Asp Arg Gly Tyr Met Asp Lys Asp Gly Tyr  
1 5 10

<210> 684

<211> 14

<212> PRT

<213> Homo sapiens

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<221> VARIANT

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<400> 684

Ser Gly Tyr Pro Lys Met Ser Ala His Thr His Ser Ser Phe  
1 5 10

<210> 685

<211> 14

<212> PRT

<213> Homo sapiens

<220>

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<222> (7)...(0)

<223> cSNP translation

<400> 685

Val Val Lys Ala Phe Val Ile Leu Ala Ser Gln Phe Leu Ser  
1 5 10

<210> 686

<211> 14

<212> PRT

<213> Homo sapiens

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<222> (7)...(0)

<223> cSNP translation

<400> 686

Asp Pro Leu Ile Tyr Ala Phe Arg Ser Gln Glu Met Arg Lys  
1 5 10

<210> 687

<211> 14

<212> PRT  
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<220>  
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<222> (7)...(0)  
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<400> 687  
Leu Ala Thr Leu Pro Glu Tyr Val Val Tyr Lys Pro Gln Met  
1 5 10

<210> 688  
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<212> PRT  
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<220>  
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<400> 688  
Leu Ala Pro Gln Gln Arg Val Ala Pro Gln Gln Lys Arg Ser  
1 5 10

<210> 689  
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<400> 689  
Leu Tyr Arg Asp Ile Phe Glu His Leu Arg Asp Glu Ser Gly  
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<210> 690  
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<400> 690  
Asp Phe Tyr Tyr Leu Gly Ala Phe Phe Gly Gly Ser Val Gln  
1 5 10

<210> 691  
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<400> 691  
Leu Thr Met Val Thr Leu Val Thr Leu Pro Leu Leu Phe Leu  
1 5 10

<210> 692  
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<400> 692  
Arg Ala Leu Tyr Leu Leu Ile Arg Arg Val Leu His Leu Gly  
1 5 10

<210> 693  
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Glu Glu Ala Met Asn Ala Val Tyr Ser Gly Tyr Val Tyr Thr  
1 5 10

<210> 694  
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<400> 694  
Ser Ala Ala Ser Tyr Asn Val Lys Thr Ala Tyr Arg Lys Tyr  
1 5 10

<210> 695  
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<220>  
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<400> 695  
Glu Ser Thr Thr Val Gly Ser Ser  
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<210> 696  
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<212> PRT  
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<400> 696  
Gln Gln Trp Ser Glu His His Ala Phe Leu Ser Gln Gly Ser  
1 5 10

<210> 697  
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<212> PRT  
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<400> 697  
Asp Asn Phe Ser Val Thr Glu Val Pro Phe Thr Glu Ser Ala  
1 5 10

<210> 698  
<211> 14  
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<400> 698  
Pro Gly Arg Arg Gln Arg Leu Thr Met Ala Ile Arg Thr Val  
1 5 10

<210> 699  
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<400> 699  
Leu Ala Gly Lys Val Ala Gln Val Lys Lys Asn Gly Arg Ile  
1 5 10

<210> 700  
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<212> PRT  
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<400> 700  
Leu Glu Asn Gln Lys Lys Val Arg Lys Lys Val Leu Ile

1 5 10

<210> 701  
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<400> 701  
Val Ser Asp Glu Glu Leu Asp Gln Met Leu Asp Ser Gly Gln  
1 5 10

<210> 702  
<211> 14  
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<220>  
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<400> 702  
Trp Leu Gly Phe Asn Lys Gln Arg Gly His Leu Gln Ile Ala  
1 5 10

<210> 703  
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<220>  
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<400> 703  
Glu Pro Glu Cys Arg Glu Val Phe His Arg Arg Ala Arg Ala  
1 5 10

<210> 704  
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<400> 704  
Leu Pro Cys Gly Pro Gly Val Lys Gly Arg Cys Phe Gly Pro  
1 5 10

<210> 705  
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<400> 705  
Ser Ala Met Asp Thr Arg Leu Leu Cys Cys Ala Val Ile Cys  
1 5 10

<210> 706  
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<220>  
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<400> 706  
Asn Thr Arg Leu Leu Cys His Val Met Leu Cys Leu Leu Gly  
1 5 10

<210> 707  
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<212> PRT  
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<220>  
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<400> 707  
Gln Thr Gly Asp Ser Ala Ile Tyr Leu Cys Ala Val Glu Ala  
1 5 10

<210> 708  
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<400> 708  
Val Tyr Leu Cys Ala Val Asp Ala Tyr Ser Asn Asp Tyr Lys  
1 5 10

<210> 709  
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<220>  
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<400> 709  
Gln Lys Gln Met Glu Leu Asp Ser Ile Leu Val Ala Leu Leu  
1 5 10

<210> 710  
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<220>  
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<400> 710  
Gly Ser Thr Val Ile Ala Gly Ser Ile Asn Ala His Gly Ser  
1 5 10

<210> 711  
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<212> PRT  
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<220>  
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<400> 711  
Pro Val Met Gly Leu Met Ile Tyr Met Met Val Met Asp His  
1 5 10

<210> 712  
<211> 14  
<212> PRT  
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<220>  
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<400> 712  
Ser Ser Gln Asp Pro Ala Ser Val Arg Glu Cys His Asp Pro  
1 5 10

<210> 713  
<211> 14  
<212> PRT  
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<220>  
<221> VARIANT  
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<400> 713  
Val Leu Leu Leu Leu Gly Ala Cys Ala Ala Pro Pro Ala Trp  
1 5 10

<210> 714  
<211> 14  
<212> PRT  
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<400> 714

Ser Leu Pro Tyr Ala Val Ala Pro Leu Ser Leu Pro Arg Gly  
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<210> 715

<211> 14

<212> PRT

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<400> 715

Tyr Gly Pro Gln Cys Gln Leu Val Ile Gln Cys Glu Pro Leu  
1 5 10

<210> 716

<211> 14

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<400> 716

Thr Met Asp Cys Thr His Ser Leu Gly Asn Phe Ser Phe Ser  
1 5 10

<210> 717

<211> 14

<212> PRT

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<400> 717

Gly Thr Thr Glu Thr Gly Gly Gln Gly Lys Gly Thr Ser Lys  
1 5 10

<210> 718

<211> 14

<212> PRT

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<220>

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<222> (7)...(0)

<223> cSNP translation

<400> 718

Cys Asn Gly Val Ala Val Cys Ser Asn Gln Asp Leu Ile Thr  
1 5 10

<210> 719

<211> 14

<212> PRT  
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<220>  
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<222> (7)...(0)  
<223> cSNP translation

<400> 719  
Leu Val Ser Tyr Cys Pro Arg Arg Leu Gln Gln Leu Leu Pro  
1 5 10

<210> 720  
<211> 14  
<212> PRT  
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<220>  
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<223> cSNP translation

<400> 720  
Arg Pro Gly Ser Pro Glu Gly Pro Leu Gly Pro Gly Gly Pro  
1 5 10

<210> 721  
<211> 14  
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<220>  
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<400> 721  
Tyr Ala Glu Arg Tyr Gln Met Pro Thr Gly Ile Lys Gly Pro  
1 5 10

<210> 722  
<211> 14  
<212> PRT  
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<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 722  
Asn Pro Leu Val Pro Gly Thr Pro Gly Arg Pro Gly Ile Pro  
1 5 10

<210> 723  
<211> 14  
<212> PRT  
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<220>  
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<400> 723  
Val Thr Asn Arg Pro Cys Gly Ser Gln Val Arg Cys Glu Gly  
1 5 10

<210> 724  
<211> 14  
<212> PRT  
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<220>  
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<222> (7)...(0)  
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<400> 724  
Ser Thr Thr Cys Val Arg Gln Ala Gln Cys Gly Gln Asp Phe  
1 5 10

<210> 725  
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<220>  
<221> VARIANT  
<222> (7)...(0)  
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<400> 725  
Gly Gly Gln Pro Cys Thr Ala Pro Leu Val Ala Phe Gln Pro  
1 5 10

<210> 726  
<211> 14  
<212> PRT  
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<220>  
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<223> cSNP translation

<400> 726  
Ser Pro Arg Ile Ser Ser Pro Arg Pro Gln Gly Leu Ser Asn  
1 5 10

<210> 727  
<211> 14  
<212> PRT  
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<220>  
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<223> cSNP translation

<400> 727  
Pro Met Thr Gln Thr Thr Ser Leu Lys Thr Ser Trp Val Asn  
1 5 10

<210> 728  
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<212> PRT  
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<220>  
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<400> 728  
Ile Asn Ala Tyr Ile Ser His Leu Gly Phe Arg Phe  
1 5 10

<210> 729  
<211> 14  
<212> PRT  
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<220>  
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<223> cSNP translation

<400> 729  
Leu His Ser Gly His Arg Gln Arg Pro Glu Phe Arg Pro Tyr  
1 5 10

<210> 730  
<211> 14  
<212> PRT  
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<400> 730  
Cys Ser Gln Glu Ala Lys Gln Ser Ala Tyr Cys Pro Tyr Ser  
1 5 10

<210> 731  
<211> 14  
<212> PRT  
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<220>  
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<223> cSNP translation

<400> 731  
Arg Thr Ile Leu Ile Phe Gly Arg Cys Gln Glu Gln Phe Gly  
1 5 10

<210> 732  
<211> 14  
<212> PRT  
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<220>  
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<400> 732  
Asn Ala Asp Ile Ile Glu Ala Leu Arg Lys Lys Gly Phe Lys

1 5 10

<210> 733  
<211> 9  
<212> PRT  
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<220>  
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<223> cSNP translation

<400> 733  
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<210> 734  
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<220>  
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<400> 734  
Ile Gln Thr Ala Glu Leu Thr Pro Glu Leu Val Ile Ser Asn  
1 5 10

<210> 735  
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<212> PRT  
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<220>  
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<400> 735  
Glu Gln Gln Gln Glu Gln His Gln Glu Gln Gln Glu Gln  
1 5 10

<210> 736  
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<400> 736  
Leu Ser Phe Ile Ser Thr Glu Leu Lys Tyr Pro Gly Met Pro  
1 5 10

<210> 737  
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<400> 737  
Ile Leu Pro Ser Gly Leu Ala Phe Ile Ser Thr Gly Leu Lys  
1 5 10

<210> 738  
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<400> 738  
Phe Tyr Leu Ala Met Asn Glu Glu Gly Lys Leu Tyr Ala Lys  
1 5 10

<210> 739  
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<400> 739  
Gly Leu Asp Gln Lys Arg Ile Lys Tyr Val Val Gly Glu Leu  
1 5 10

<210> 740  
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<220>  
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1 5 10

<210> 741  
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<400> 741  
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1 5 10

<210> 742  
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<220>  
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<400> 742  
Gly Gln Lys Thr Leu Thr Pro Val Gly Tyr Gln Ser Val Thr  
1 5 10

<210> 743  
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<400> 743  
Thr Glu Ile Thr Gly Ala Thr Thr Met Thr Ser Val Gly His  
1 5 10

<210> 744  
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<400> 744  
Gly Val Tyr Ile Leu Thr Tyr Asn Thr Ser Gln Tyr Asp Thr  
1 5 10

<210> 745  
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<400> 745  
Pro Leu Ser Phe His Val Ile Trp Ile Ala Ser Phe Tyr Asn  
1 5 10

<210> 746  
<211> 14  
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<220>  
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<223> cSNP translation

<400> 746  
Ser Thr Ile Val Phe Leu Cys Pro Trp Ser Arg Gly Asn Phe  
1 5 10

<210> 747  
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<400> 747  
Pro Asp Pro Arg Glu Ala Cys Gly Ser Ser Ser Tyr Val  
1 5 10

<210> 748  
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<400> 748  
Ala Lys Asp Met Asn Gly Thr Ser Leu His Gly Lys Ala Ile  
1 5 10

<210> 749  
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<220>  
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<222> (7)...(0)  
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<400> 749  
Arg Ser Ala Arg Gly Ser Arg Gly Gly Thr Arg Gly Trp Leu  
1 5 10

<210> 750  
<211> 14  
<212> PRT  
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<220>  
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<400> 750  
Asn Phe Thr Ser Lys Tyr His Met Lys Val Leu Tyr Leu Ser  
1 5 10

<210> 751

<211> 14  
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<220>  
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<222> (7)...(0)  
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<400> 751  
Ile Gln Tyr Thr Tyr Leu Gly Gly His Val Cys Leu Ser Ala  
1 5 10

<210> 752  
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<212> PRT  
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<400> 752  
Leu Asp Pro Asn Asn Pro Asp Ala Asn Trp Ile His Ala Arg  
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<210> 753  
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<212> PRT  
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<220>  
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<400> 753  
Cys Leu Thr Glu Arg Gln Ser Lys Ile Trp Phe Gln Asn Arg  
1 5 10

<210> 754  
<211> 14  
<212> PRT  
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<220>  
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<222> (7)...(0)  
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<400> 754  
Pro Tyr Arg Ile Ala His Ala Val Ile Lys Ala His Ala Arg  
1 5 10

<210> 755  
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<212> PRT  
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<400> 755  
Lys Arg Lys Lys Glu Val His Ala Thr Ser Pro Ala Pro Ser  
1 5 10

<210> 756  
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<220>  
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1 5 10

<210> 757  
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1 5 10

<210> 758  
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<220>  
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<400> 758  
Phe Ser Tyr Ser Ala Ser Ser Thr  
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<210> 759  
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<212> PRT  
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<400> 759  
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1 5 10

<210> 760  
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<400> 760

Trp Glu Arg Phe Val His Arg Glu Asn Gln His Leu Val Ser  
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<210> 761

<211> 14

<212> PRT

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<400> 761

Gly Thr Glu Asp Leu Tyr Gly Tyr Ile Asp Lys Tyr Asn Ile  
1 5 10

<210> 762

<211> 14

<212> PRT

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<400> 762

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<210> 763

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<212> PRT

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<400> 763

Gly Ser Leu His Pro His Pro Pro Tyr His Ile Arg Val Ala  
1 5 10

<210> 764

<211> 14

<212> PRT

<213> Homo sapiens

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<400> 764

Leu Tyr Ser Arg Leu Gly Gly Gln Pro Val Tyr Leu Pro Thr  
1 5 10

<210> 765  
<211> 14  
<212> PRT  
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<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 765  
Ala Pro Pro Gly Ala Tyr His Gly Ala Pro Gly Ala Tyr Pro  
1 5 10

<210> 766  
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<212> PRT  
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<220>  
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<400> 766  
Val Met Pro His Ser Ser Glu His Lys Thr Ala Gln Pro Asn  
1 5 10

<210> 767  
<211> 14  
<212> PRT  
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<220>  
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<400> 767  
Thr Cys Val Val Glu His Thr Gly Ala Pro Glu Pro Ile Leu  
1 5 10

<210> 768  
<211> 14  
<212> PRT  
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<220>  
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<400> 768  
Val Gly Phe Leu Val Gly Ile Val Leu Ile Ile Met Gly Thr  
1 5 10

<210> 769  
<211> 14  
<212> PRT  
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<220>  
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<400> 769  
Gln Ser Val Val Ser Cys Ala  
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<210> 770  
<211> 14  
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<223> cSNP translation

<400> 770  
Pro Gly Pro Thr Val Arg Ala Gly Glu Asn Val Thr Leu Ser  
1 5 10

<210> 771  
<211> 14  
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<400> 771  
Cys Ser Gly Val Trp Gly Ala Asp Thr Glu Glu Arg Leu Val  
1 5 10

<210> 772  
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<400> 772  
Ala Leu Leu Arg His Glu Leu Lys Gly Tyr Gln Lys Trp Val  
1 5 10

<210> 773  
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<400> 773  
Leu Leu Arg His Glu Trp Gln Gly Tyr Gln Lys Trp Val Arg  
1 5 10

<210> 774  
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<400> 774  
Gly Phe His Tyr Gly Val Leu Ala Cys Glu Gly Cys Lys Gly  
1 5 10

<210> 775  
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<400> 775  
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1 5 10

<210> 776  
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<400> 776  
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<220>  
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<400> 777  
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<210> 778  
<211> 14  
<212> PRT  
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<220>  
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<222> (7)...(0)  
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<400> 778  
Leu Val Val Val Gly Ala Cys Gly Val Gly Lys Ser Ala Leu  
1 5 10

<210> 779  
<211> 14  
<212> PRT  
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<400> 779  
Ile Leu Asp Thr Ala Gly His Glu Glu Tyr Ser Ala Met Arg  
1 5 10

<210> 780  
<211> 14  
<212> PRT  
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<220>  
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<222> (7)...(0)  
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<400> 780  
Phe Gly His Gln Glu Asn Ser Gln Asn Glu Glu Ile Leu Asn  
1 5 10

<210> 781  
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<212> PRT  
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<400> 781  
Leu Thr Tyr Pro Pro Gly Pro Arg Thr Gln Leu Arg Glu Asp  
1 5 10

<210> 782  
<211> 14  
<212> PRT  
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<220>  
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<400> 782  
Gln Pro Gln Ala Arg Gln Glu Glu Gln Val Arg Val Val Arg  
1 5 10

<210> 783

<211> 14  
<212> PRT  
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<220>  
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<400> 783  
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1 5 10

<210> 784  
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<212> PRT  
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<220>  
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<400> 784  
Ala Gln Ile Phe Asp Tyr Ser Glu Ile Pro Asn Phe Pro Arg  
1 5 10

<210> 785  
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<212> PRT  
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<220>  
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<400> 785  
Asp Asp Asp Asp Ala Pro Arg Pro Ser Gln Phe Glu Glu Asp  
1 5 10

<210> 786  
<211> 14  
<212> PRT  
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<220>  
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<400> 786  
His Val Leu Arg Met His Gly Tyr Arg Ala Pro Gly Glu Gln  
1 5 10

<210> 787  
<211> 14  
<212> PRT  
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<220>  
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<400> 787  
Glu Thr Gln Leu Gly Thr Leu Ala Gln Phe Pro Asn Thr Leu  
1 5 10

<210> 788  
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<212> PRT  
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<220>  
<221> VARIANT  
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<400> 788  
Glu Ala Met Glu Arg Phe Gly Glu Asp Glu Gly Phe Ile Lys  
1 5 10

<210> 789  
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<212> PRT  
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<220>  
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<400> 789  
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1 5 10

<210> 790  
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<212> PRT  
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<220>  
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1 5 10

<210> 791  
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<212> PRT  
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<220>  
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<222> (7)...(0)  
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<400> 791  
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<210> 792  
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<213> Homo sapiens

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5

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<210> 793

<211> 14

<212> PRT

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<221> VARIANT

<222> (7)...(0)

<223> cSNP translation

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Glu Ala Glu Arg Val Lys Glu Gln Thr Phe Arg Glu Lys Ala

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<210> 794

<211> 14

<212> PRT

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<221> VARIANT

<222> (7)...(0)

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Ala Phe Val Val Leu Ala Leu Gln Phe Leu Ser His Asp Pro

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<210> 795

<211> 14

<212> PRT

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<222> (7)...(0)

<223> cSNP translation

<400> 795

Thr Gly Lys Ile Gln Arg Thr Lys Leu Arg Asp Lys Glu Trp

1

5

10

<210> 796

<211> 14

<212> PRT

<213> Homo sapiens

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<222> (7)...(0)

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<400> 796

Val Asp Tyr Met Thr Gln Ala Arg Gly Gln Arg Ser Ser Leu  
1 5 10

<210> 797  
<211> 14  
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<220>  
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1 5 10

<210> 798  
<211> 14  
<212> PRT  
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<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 798  
Leu Arg Ile Gln Cys Leu Cys Arg Lys Gln Ser Ser Lys His  
1 5 10

<210> 799  
<211> 14  
<212> PRT  
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<220>  
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<222> (7)...(0)  
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<400> 799  
Leu Glu Lys Ile Gln Pro Met Thr Gln Asn Gly Gln His Pro  
1 5 10

<210> 800  
<211> 14  
<212> PRT  
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<220>  
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<400> 800  
Trp Met Ile Phe Val Val Ile Ala Ser Val Phe Thr Asn Gly  
1 5 10

<210> 801  
<211> 14  
<212> PRT  
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<220>  
<221> VARIANT  
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<223> cSNP translation

<400> 801  
Ser Ile Asn Leu Phe Ser Gly Ile Phe Phe Leu Thr Cys Met  
1 5 10

<210> 802  
<211> 14  
<212> PRT  
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<220>  
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<222> (7)...(0)  
<223> cSNP translation

<400> 802  
Val Asp Ile Cys Phe Ser Ser Thr Thr Val Pro Lys Met Leu  
1 5 10

<210> 803  
<211> 14  
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<220>  
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<223> cSNP translation

<400> 803  
Glu Ser Asn Thr Thr Gly Thr Thr Ala Phe Ser Met Pro Ser  
1 5 10

<210> 804  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 804  
Val His Pro Val Arg Pro Leu Arg Leu Glu Ser Phe Ser Ala  
1 5 10

<210> 805  
<211> 14  
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<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 805  
Arg Gly Ala Arg Pro Gly Pro Arg Val Pro Lys Thr Leu Val  
1 5 10

&lt;210&gt; 806

&lt;211&gt; 14

&lt;212&gt; PRT

&lt;213&gt; Homo sapiens

&lt;220&gt;

&lt;221&gt; VARIANT

&lt;222&gt; (7)...(0)

&lt;223&gt; cSNP translation

&lt;400&gt; 806

Lys Ala Phe Leu Thr Ser His Ser Leu Arg Ile His Val Arg  
1 5 10

&lt;210&gt; 807

&lt;211&gt; 14

&lt;212&gt; PRT

&lt;213&gt; Homo sapiens

&lt;220&gt;

&lt;221&gt; VARIANT

&lt;222&gt; (7)...(0)

&lt;223&gt; cSNP translation

&lt;400&gt; 807

Ser Glu Ala Ser Ser Ala Phe Phe Met Ala Lys Lys Lys Thr  
1 5 10

&lt;210&gt; 808

&lt;211&gt; 14

&lt;212&gt; PRT

&lt;213&gt; Homo sapiens

&lt;220&gt;

&lt;221&gt; VARIANT

&lt;222&gt; (7)...(0)

&lt;223&gt; cSNP translation

&lt;400&gt; 808

Glu Leu Tyr Arg Asp Ile Leu Gln His Leu Arg Asp Glu Ser  
1 5 10

&lt;210&gt; 809

&lt;211&gt; 14

&lt;212&gt; PRT

&lt;213&gt; Homo sapiens

&lt;220&gt;

&lt;221&gt; VARIANT

&lt;222&gt; (7)...(0)

&lt;223&gt; cSNP translation

&lt;400&gt; 809

Tyr Val Phe Thr Asp Gln Leu Ala Ala Val Pro Arg Val Thr  
1 5 10

&lt;210&gt; 810

&lt;211&gt; 14

&lt;212&gt; PRT

&lt;213&gt; Homo sapiens

&lt;220&gt;

&lt;221&gt; VARIANT

<222> (7)...(0)  
<223> cSNP translation

<400> 810  
Leu Ser Val Leu Glu Val Gly Ala Tyr Lys Arg Trp Gln Asp  
1 5 10

<210> 811  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
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<223> cSNP translation

<400> 811  
Val Asp Val Asp Met Glu Ile Arg Asp His Val Gly Val Glu  
1 5 10

<210> 812  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 812  
Phe Gly Thr Leu His Pro Ser Phe Tyr Gly Ser Ser Arg Glu  
1 5 10

<210> 813  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 813  
Glu Gly Asp Phe Tyr Tyr Met Gly Gly Phe Phe Gly Gly Ser  
1 5 10

<210> 814  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 814  
Gly Gly Ser Val Gln Gln Glu Met Gln Arg Leu Thr Arg Ala Cys  
1 5 10

<210> 815

&lt;211&gt; 14

&lt;212&gt; PRT

&lt;213&gt; Homo sapiens

&lt;220&gt;

&lt;221&gt; VARIANT

&lt;222&gt; (7)...(0)

&lt;223&gt; cSNP translation

&lt;400&gt; 815

Val Val Glu Leu Thr Gln Asp Asp Ala Leu Gly Ser Arg Trp  
1 5 10

&lt;210&gt; 816

&lt;211&gt; 14

&lt;212&gt; PRT

&lt;213&gt; Homo sapiens

&lt;220&gt;

&lt;221&gt; VARIANT

&lt;222&gt; (7)...(0)

&lt;223&gt; cSNP translation

&lt;400&gt; 816

Lys Asp Phe His Lys Asp Met Leu Lys Pro Ser Pro Gly Lys  
1 5 10

&lt;210&gt; 817

&lt;211&gt; 14

&lt;212&gt; PRT

&lt;213&gt; Homo sapiens

&lt;220&gt;

&lt;221&gt; VARIANT

&lt;222&gt; (7)...(0)

&lt;223&gt; cSNP translation

&lt;400&gt; 817

Thr Asn Asn Cys Tyr Arg His Ala Ile Val Thr Thr Ser Ile  
1 5 10

&lt;210&gt; 818

&lt;211&gt; 14

&lt;212&gt; PRT

&lt;213&gt; Homo sapiens

&lt;220&gt;

&lt;221&gt; VARIANT

&lt;222&gt; (7)...(0)

&lt;223&gt; cSNP translation

&lt;400&gt; 818

Gly Phe Val Val Phe Ser Ser Leu Gly Tyr Met Ala Gln Lys  
1 5 10

&lt;210&gt; 819

&lt;211&gt; 14

&lt;212&gt; PRT

&lt;213&gt; Homo sapiens

&lt;220&gt;

&lt;221&gt; VARIANT

&lt;222&gt; (7)...(0)

&lt;223&gt; cSNP translation

<400> 819  
Met Asp Glu Ala Ala Arg Pro Glu Ala Trp Asp Ser Tyr Arg  
1 5 10

<210> 820  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 820  
Ser Pro Gln Ser Ser Ala Arg Gly Lys Pro Ala Met Ser Tyr  
1 5 10

<210> 821  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
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<222> (7)...(0)  
<223> cSNP translation

<400> 821  
Leu Glu Asp Leu Ala Gly Trp Lys Glu Leu Phe Gln Thr Pro  
1 5 10

<210> 822  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
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<222> (7)...(0)  
<223> cSNP translation

<400> 822  
Val Gln Asp Ile Leu Arg Leu Glu Met Pro Ala Ser Lys Ile  
1 5 10

<210> 823  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
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<222> (7)...(0)  
<223> cSNP translation

<400> 823  
Ser Glu Arg Glu Thr Glu His Thr Pro Ala Leu Ile Met Val  
1 5 10

<210> 824  
<211> 14  
<212> PRT

<213> Homo sapiens

<220>

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<222> (7)...(0)

<223> cSNP translation

<400> 824

Lys Val Leu Asp His Trp Cys Ile Met Thr Ser Glu Glu Glu

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<210> 825

<211> 14

<212> PRT

<213> Homo sapiens

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<222> (7)...(0)

<223> cSNP translation

<400> 825

Gln Glu Val His Gly Pro Tyr Pro Asp Ser Ser Phe Leu Thr

1

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<210> 826

<211> 14

<212> PRT

<213> Homo sapiens

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<221> VARIANT

<222> (7)...(0)

<223> cSNP translation

<400> 826

Gln Glu Val His Gly Pro Ile Pro Asp Ser Ser Phe Leu Thr

1

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<210> 827

<211> 14

<212> PRT

<213> Homo sapiens

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<221> VARIANT

<222> (7)...(0)

<223> cSNP translation

<400> 827

Glu Leu Cys His Glu Lys Gly Ile Leu Glu Lys Tyr Gly His

1

5

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<210> 828

<211> 14

<212> PRT

<213> Homo sapiens

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<221> VARIANT

<222> (7)...(0)

<223> cSNP translation

<400> 828

Asn Asn Asn Leu Arg His Thr Asp Glu Met Phe Trp Asn His  
1 5 10

<210> 829  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 829  
Gly Met Ala Ser Ser Cys Ser Val Gln Val Lys Leu Glu Leu  
1 5 10

<210> 830  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 830  
Pro Ser Ile Phe Ile Tyr Arg His Thr Ala Ser Gly Lys Thr  
1 5 10

<210> 831  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 831  
Pro Pro Pro Pro Pro Gly Ala Pro Gly Gly Ser Gln Asp Thr  
1 5 10

<210> 832  
<211> 14  
<212> PRT  
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<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 832  
Arg Ala Ala Leu Glu Arg Gly Lys Ala Ile Glu Lys Asn Leu  
1 5 10

<210> 833  
<211> 14  
<212> PRT  
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<220>  
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<400> 833  
Thr Gly Gly Leu Leu Leu Arg Leu Ala Leu Met Leu Gln Leu  
1 5 10

<210> 834  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
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<400> 834  
Asp Ser Ser Ser Ser Asn Gly Lys Ala Lys Asn Pro Pro Gly  
1 5 10

<210> 835  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
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<222> (7)...(0)  
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<400> 835  
Leu Glu Pro Gln Trp Tyr Ser Val Leu Glu Lys Asp Ser Val  
1 5 10

<210> 836  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 836  
Gln Lys His Ser Ser Gly Xaa Ser Asn Thr Ser Thr Ala Asn  
1 5 10

<210> 837  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
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<222> (7)...(0)  
<223> cSNP translation

<400> 837  
Trp Gly Thr Glu Asp Asp Ala Thr Gln Ser Tyr His Asn Gly  
1 5 10

&lt;210&gt; 838

&lt;211&gt; 14

&lt;212&gt; PRT

&lt;213&gt; Homo sapiens

&lt;220&gt;

&lt;221&gt; VARIANT

&lt;222&gt; (7)...(0)

&lt;223&gt; cSNP translation

&lt;400&gt; 838

Arg Cys Met Gly Thr Val Asn Leu Asn Gln Ala Arg Gly Ser  
1 5 10

&lt;210&gt; 839

&lt;211&gt; 14

&lt;212&gt; PRT

&lt;213&gt; Homo sapiens

&lt;220&gt;

&lt;221&gt; VARIANT

&lt;222&gt; (7)...(0)

&lt;223&gt; cSNP translation

&lt;400&gt; 839

Phe Ser Lys Leu Leu Gly Pro Leu Ser Ala Lys Lys Tyr Leu  
1 5 10

&lt;210&gt; 840

&lt;211&gt; 14

&lt;212&gt; PRT

&lt;213&gt; Homo sapiens

&lt;220&gt;

&lt;221&gt; VARIANT

&lt;222&gt; (7)...(0)

&lt;223&gt; cSNP translation

&lt;400&gt; 840

Thr Ser Lys Ile Leu Phe Phe Ser Gln Gly Ser Glu Ile Ala  
1 5 10

&lt;210&gt; 841

&lt;211&gt; 14

&lt;212&gt; PRT

&lt;213&gt; Homo sapiens

&lt;220&gt;

&lt;221&gt; VARIANT

&lt;222&gt; (7)...(0)

&lt;223&gt; cSNP translation

&lt;400&gt; 841

Tyr Val Gln Pro Pro Glu Val Ile Gly Pro Met Arg Pro Glu  
1 5 10

&lt;210&gt; 842

&lt;211&gt; 14

&lt;212&gt; PRT

&lt;213&gt; Homo sapiens

&lt;220&gt;

&lt;221&gt; VARIANT

<222> (7)...(0)  
<223> cSNP translation

<400> 842  
Gly Pro Asp Gly Gln Glu Val Asp Pro Pro Asn Pro Glu Glu  
1 5 10

<210> 843  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 843  
Lys Gly Gly Glu Gln Lys Arg His Glu Lys Ile Ser Ala Ser  
1 5 10

<210> 844  
<211> 14  
<212> PRT  
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<220>  
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<222> (7)...(0)  
<223> cSNP translation

<400> 844  
Ala Pro Gln Gln Glu Gly Glu Ala Ser Lys Glu Lys Glu Glu  
1 5 10

<210> 845  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
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<222> (7)...(0)  
<223> cSNP translation

<400> 845  
Lys Val Val Gln Ser Pro Gln Ser Leu Val Val His Glu Gly  
1 5 10

<210> 846  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 846  
Leu Asn Cys Ser Tyr Glu Met Thr Asn Phe Arg Ser Leu Leu  
1 5 10

<210> 847

<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 847  
Thr Asn Phe Arg Ser Leu Gln Trp Tyr Lys Gln Glu Lys Lys  
1 5 10

<210> 848  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 848  
Leu Asp Lys Lys Glu Leu Ser Ser Ile Leu Asn Ile Thr Ala  
1 5 10

<210> 849  
<211> 14  
<212> PRT  
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<220>  
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<223> cSNP translation

<400> 849  
Glu Tyr Val Val Gly Ala Pro His Leu Glu Leu Asp Pro Gly  
1 5 10

<210> 850  
<211> 14  
<212> PRT  
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<220>  
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<223> cSNP translation

<400> 850  
Phe Phe Lys Arg Asn Arg His Thr Pro Gly Arg Arg  
1 5 10

<210> 851  
<211> 9  
<212> PRT  
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<220>  
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<223> cSNP translation

<400> 851  
Thr Ile Gln Pro Pro Arg Glu  
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<210> 852  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (8)...(0)  
<223> cSNP translation

<400> 852  
Gly Ile Val Gly Gln Lys Gly Arg Pro Trp Leu Pro Arg Thr  
1 5 10

<210> 853  
<211> 14  
<212> PRT  
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<220>  
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<222> (8)...(0)  
<223> cSNP translation

<400> 853  
Gly Gly Lys Met Gly Gly Arg Lys Arg Leu Gln Lys  
1 5 10

<210> 854  
<211> 14  
<212> PRT  
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<220>  
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<400> 854  
Tyr Ser Ser Tyr Gly Gln Ser Leu Phe Thr Val Leu Trp Trp  
1 5 10

<210> 855  
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<220>  
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<400> 855  
Glu Gln Leu Arg Arg Gln Leu Asp Pro Leu Arg Thr Ala His  
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<210> 856  
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<213> Homo sapiens

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<222> (7)...(0)

<223> cSNP translation

<400> 856

Ser Thr Glu Cys Trp Met Asn Ala Ala Cys Leu Ala Pro Gly  
1 5 10

<210> 857

<211> 14

<212> PRT

<213> Homo sapiens

<220>

<221> VARIANT

<222> (9)...(0)

<223> cSNP translation

<400> 857

His Gly Val Leu Asp Ala Cys Leu Ile His Pro Gly Pro Ala  
1 5 10

<210> 858

<211> 14

<212> PRT

<213> Homo sapiens

<220>

<221> VARIANT

<222> (7)...(0)

<223> cSNP translation

<400> 858

Arg Asp Lys Gly Ser Gly Arg Ala Cys Gly Leu Glu Gly Gln  
1 5 10

<210> 859

<211> 14

<212> PRT

<213> Homo sapiens

<220>

<221> VARIANT

<222> (7)...(0)

<223> cSNP translation

<400> 859

Thr Asp Phe Phe Phe Phe  
1 5

<210> 860

<211> 14

<212> PRT

<213> Homo sapiens

<220>

<221> VARIANT

<222> (8)...(0)

<223> cSNP translation

<400> 860

Gly Ala Gly Ser Val Ser Asp His His Ser Ile Thr Lys  
1 5 10

<210> 861

<211> 12

<212> PRT

<213> Homo sapiens

<220>

<221> VARIANT

<222> (7)...(0)

<223> cSNP translation

<400> 861

Arg Tyr Leu Asp Trp Ile Leu Trp Ala His Gln Arg  
1 5 10

<210> 862

<211> 14

<212> PRT

<213> Homo sapiens

<220>

<221> VARIANT

<222> (8)...(0)

<223> cSNP translation

<400> 862

Ala Glu Leu Arg Leu Leu Arg Ala Gln Val Lys Ser Gly Ala  
1 5 10

<210> 863

<211> 14

<212> PRT

<213> Homo sapiens

<220>

<221> VARIANT

<222> (8)...(0)

<223> cSNP translation

<400> 863

Ala Glu Leu Arg Leu Leu Arg Ala Gln Val Lys Ser Gly Ala  
1 5 10

<210> 864

<211> 14

<212> PRT

<213> Homo sapiens

<220>

<221> VARIANT

<222> (8)...(0)

<223> cSNP translation

<400> 864

Pro His Cys Arg Pro Gly Ala Trp Pro Ala Thr Glu Arg Gly  
1 5 10

<210> 865

<211> 14

<212> PRT

<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (8)...(0)  
<223> cSNP translation

<400> 865  
Ile His Phe Glu Asp Tyr Gly Val Leu Gly His His Gln Leu  
1 5 10

<210> 866  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
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<222> (7)...(0)  
<223> cSNP translation

<400> 866  
Asn Phe Ile Leu Ala Cys Pro Arg  
1 5

<210> 867  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 867  
Arg Glu Lys Leu Arg Asn Phe His Phe Ser Met Ser Arg  
1 5 10

<210> 868  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (8)...(0)  
<223> cSNP translation

<400> 868  
Leu Leu Leu Leu Leu Leu Arg Arg Pro Ala Gln Pro Gln Leu  
1 5 10

<210> 869  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 869  
Lys Arg Val Ala Gly Gly Leu Arg  
1 5

<210> 870  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (8)...(0)  
<223> cSNP translation

<400> 870  
Gly Lys Arg Val Ala Gly Gly Leu Arg  
1 5

<210> 871  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 871  
Ser Ser Gly Arg Pro Thr Gly Tyr Cys Leu Gln Leu Gln Gln  
1 5 10

<210> 872  
<211> 14  
<212> PRT  
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<220>  
<221> VARIANT  
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<223> cSNP translation

<400> 872  
Gln Arg Ser Ile Ser Ala Asp  
1 5

<210> 873  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (8)...(0)  
<223> cSNP translation

<400> 873  
Arg Ala Pro Val Ile Leu Gly Pro Pro Thr Thr Cys Ser Ser  
1 5 10

<210> 874  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT

<222> (7)...(0)  
<223> cSNP translation

<400> 874  
Met Gly Leu Ser Gly Phe Leu Thr Gly Pro Pro Pro Pro Gly  
1 5 10

<210> 875  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (8)...(0)  
<223> cSNP translation

<400> 875  
Leu Met Gly Leu Ser Gly Phe Leu Thr Gly Pro Pro Pro Pro  
1 5 10

<210> 876  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 876  
Pro Arg Thr Pro Ala Glu Pro Pro Pro Leu Gly Arg Gln Ala  
1 5 10

<210> 877  
<211> 14  
<212> PRT  
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<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 877  
Gly Thr Gly Asp Trp Arg Glu Pro Gly Ala Ala Ser Glu Arg  
1 5 10

<210> 878  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (9)...(0)  
<223> cSNP translation

<400> 878  
Gln Gly Arg Gln Ser Lys Gly Leu Arg Arg Arg Thr Trp Pro  
1 5 10

<210> 879

<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (8)...(0)  
<223> cSNP translation

<400> 879  
Lys Cys Lys Cys Ser Arg Lys Asp Pro Arg Ser Ala Thr Ala  
1 5 10

<210> 880  
<211> 14  
<212> PRT  
<213> Homo sapiens

<220>  
<221> VARIANT  
<222> (7)...(0)  
<223> cSNP translation

<400> 880  
Cys Lys Cys Ser Arg Lys Asp Pro Arg Ser Ala Thr Ala Thr  
1 5 10

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